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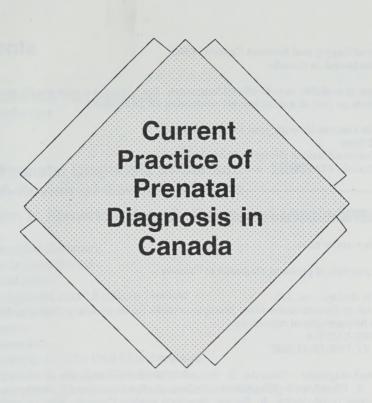
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CURRENT PRACTICE OF PRENATAL DIAGNOSIS IN CANADA

Research Studies of the Royal Commission on New Reproductive Technologies





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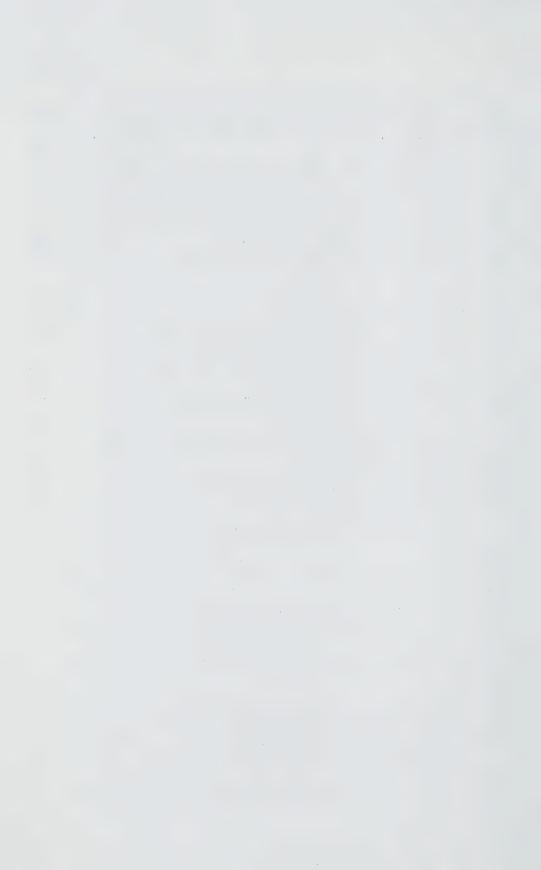
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Preface from the Chairperson



As Canadians living in the last decade of the twentieth century, we face unprecedented choices about procreation. Our responses to those choices — as individuals and as a society — say much about what we value and what our priorities are. Some technologies, such as those for assisted reproduction, are unlikely to become a common means of having a family — although the number of children born as a result of these techniques is greater than the number of infants placed for adoption in Canada. Others, such as ultrasound during pregnancy, are already generally accepted, and half of all pregnant women aged 35 and over undergo prenatal diagnostic procedures. Still other technologies, such as fetal tissue research, have little to do with reproduction as such, but may be of benefit to people suffering from diseases such as Parkinson's; they raise important ethical issues in the use and handling of reproductive tissues.

It is clear that opportunities for technological intervention raise issues that affect all of society; in addition, access to the technologies depends on the existence of public structures and policies to provide them. The values and priorities of society, as expressed through its institutions, laws, and funding arrangements, will affect individual options and choices.

As Canadians became more aware of these technologies throughout the 1980s, there was a growing awareness that there was an unacceptably large gap between the rapid pace of technological change and the policy development needed to guide decisions about whether and how to use such powerful technologies. There was also a realization of how little reliable information was available to make the needed policy decisions. In addition, many of the attitudes and assumptions underlying the way in which technologies were being developed and made available did not reflect the profound changes that have been transforming Canada in recent decades. Individual cases were being dealt with in isolation, and often in the absence of informed social consensus. At the same time, Canadians were looking

more critically at the role of science and technology in their lives in general, becoming more aware of their limited capacity to solve society's problems.

These concerns came together in the creation of the Royal Commission on New Reproductive Technologies. The Commission was established by the federal government in October 1989, with a wide-ranging and complex mandate. It is important to understand that the Commission was asked to consider the technologies' impact not only on society, but also on specific groups in society, particularly women and children. It was asked to consider not only the technologies' scientific and medical aspects, but also their ethical, legal, social, economic, and health implications. Its mandate was extensive, as it was directed to examine not only current developments in the area of new reproductive technologies, but also potential ones; not only techniques related to assisted conception, but also those of prenatal diagnosis; not only the condition of infertility, but also its causes and prevention; not only applications of technology, but also research, particularly embryo and fetal tissue research.

The appointment of a Royal Commission provided an opportunity to collect much-needed information, to foster public awareness and public debate, and to provide a principled framework for Canadian public policy on the use or restriction of these technologies.

The Commission set three broad goals for its work: to provide direction for public policy by making sound, practical, and principled recommendations; to leave a legacy of increased knowledge to benefit Canadian and international experience with new reproductive technologies; and to enhance public awareness and understanding of the issues surrounding new reproductive technologies to facilitate public participation in determining the future of the technologies and their place in Canadian society.

To fulfil these goals, the Commission held extensive public consultations, including private sessions for people with personal experiences of the technologies that they did not want to discuss in a public forum, and it developed an interdisciplinary research program to ensure that its recommendations would be informed by rigorous and wide-ranging research. In fact, the Commission published some of that research in advance of the Final Report to assist those working in the field of reproductive health and new reproductive technologies and to help inform the public.

The results of the research program are presented in these volumes. In all, the Commission developed and gathered an enormous body of information and analysis on which to base its recommendations, much of it available in Canada for the first time. This solid base of research findings helped to clarify the issues and produce practical and useful recommendations based on reliable data about the reality of the situation, not on speculation.

The Commission sought the involvement of the most qualified researchers to help develop its research projects. In total, more than 300

scholars and academics representing more than 70 disciplines — including the social sciences, humanities, medicine, genetics, life sciences, law, ethics, philosophy, and theology — at some 21 Canadian universities and 13 hospitals, clinics, and other institutions were involved in the research program.

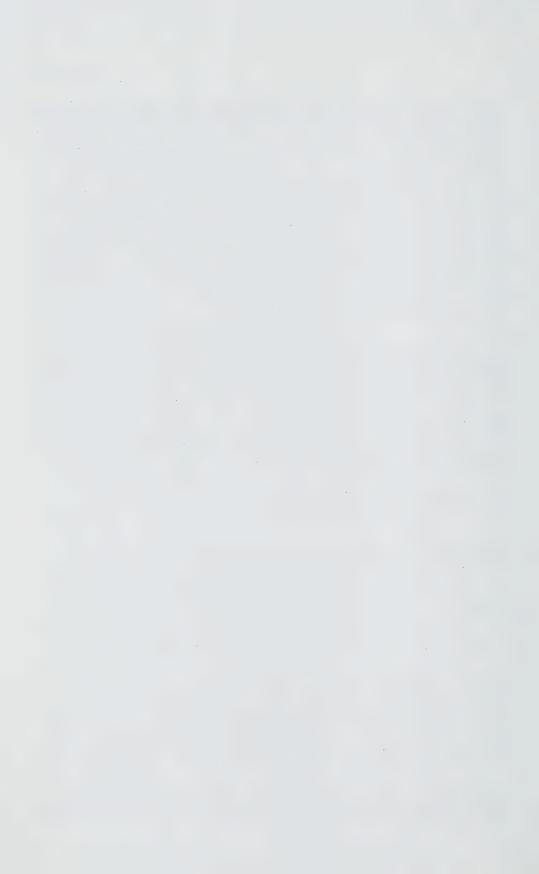
The Commission was committed to a research process with high standards and a protocol that included internal and external peer review for content and methodology, first at the design stage and later at the report stage. Authors were asked to respond to these reviews, and the process resulted in the achievement of a high standard of work. The protocol was completed before the publication of the studies in this series of research volumes. Researchers using human subjects were required to comply with appropriate ethical review standards.

These volumes of research studies reflect the Commission's wide mandate. We believe the findings and analysis contained in these volumes will be useful for many people, both in this country and elsewhere.

Along with the other Commissioners, I would like to take this opportunity to extend my appreciation and thanks to the researchers and external reviewers who have given tremendous amounts of time and thought to the Commission. I would also like to acknowledge the entire Commission staff for their hard work, dedication, and commitment over the life of the Commission. Finally, I would like to thank the more than 40 000 Canadians who were involved in the many facets of the Commission's work. Their contribution has been invaluable.

Patricia a. Baird

Patricia Baird, M.D., C.M., FRCPC, F.C.C.M.G.



Introduction



Prenatal diagnosis (PND) has become a routinely offered part of pregnancy care for a significant percentage of women in Canada — those women who are at higher risk because of their age or because of a family history, such as having had a child with a genetic disease or congenital anomaly. As outlined in Volume 12, Canada's record with regard to the introduction and provision of prenatal diagnostic techniques has been a good example internationally of responsible use of technology. This volume documents in some detail current practices in prenatal diagnosis and testing in Canada today and, in the process, provides answers to many questions raised in the Commission's public consultations.

This is done through an analysis of the activities of all Canadian genetics centres providing PND, as well as an analysis of the process by which women are referred to these centres. Other studies in the volume focus not on prenatal diagnostic procedures but on prenatal screening programs. These programs involve a wider range of physicians, often including general practitioners, and the studies examine how such programs have been introduced in Canada and the extent to which, in this wider arena, they conform to the standards established for the introduction of prenatal diagnostic techniques.

While some of these programs, such as Manitoba's maternal serum AFP screening program, involve the use of fairly new techniques, the technologies assessed in this volume are what could be termed "recognized" technologies. The next volume looks at newer and as yet unproven or speculative uses of PND and genetic technologies.

The Studies

John Hamerton, Jane Evans, and Leonie Stranc have provided the most extensive data to date on the practice of PND in Canada from a detailed survey of the country's 22 genetics centres. Their report provides

1990 data on the genetics centres, the patients referred to them, the range of services offered, the tests provided and their results, the pregnancy outcomes associated with their use, the policies governing their use, and the professionals working in these centres. In 1990, more than 20 000 women, or about 5 percent of all pregnant women, were referred to the centres for PND, the majority because they were 35 years of age or older. Just over 50 percent of pregnant women in Canada who were over age 34 were referred. The authors found that, overall, PND in Canada is being used in a "responsible, conscientious, and sensitive manner."

The survey reveals that women take into account the nature and severity of any disorder that is found in making decisions about whether to terminate a pregnancy. The finding that PND and the decisions it entails are not taken lightly by women in Canada echoes the findings of studies in Volume 12, which examine the perceptions and attitudes of women regarding PND.

The survey also reveals, however, that not all women are given the same opportunity to make decisions in this area. It finds that there is significant regional variation in referrals to genetics centres — 64 percent of women from Quebec over age 34 were referred to genetics centres; the figures for Newfoundland and Saskatchewan were 15 and 22 percent, respectively. The Commission's survey of attitudes toward new reproductive technology (in Volume 2) makes it clear that regional variations in referral rates do not reflect differences in the value that women in various regions across the country attach to PND — indeed, 84 percent of Canadians support its use. The study by Marc Renaud and colleagues on physicians' attitudes to PND, which is discussed below, sheds some additional light on the factors underlying these wide variations in referrals.

Another finding that is of interest is that, as well as a wide variation in utilization rates across the country, there is also extensive variation from centre to centre in the way information and counselling services are delivered. The stressful nature of PND, the magnitude of the decisions that have to be made should an anomaly be detected, and the importance of information as a prerequisite for the exercise of informed consent and informed choice require that patient education material be clear, understandable, and comprehensive in its provision of the content that patients need. Janis Wood Catano analyzed the readability of patient education materials used by Canada's genetics centres and found that, in general, they do not measure up to these requirements. Overall, she notes, the material is complex, technical, and difficult to read. Despite the fact that the educational level of patients at the centres is high compared with that of Canadians in general, the reading level of the informational material is still too high to be easily understood by the patients. She finds, however, some materials that are readily understandable and visually attractive, and suggests that these could form the basis for cooperative action by all

genetics centres to develop patient information materials that have a consistently high level of readability in all Canadian centres.

As noted above, Marc Renaud and colleagues examined some of the factors that may help to explain differences across the country in referral rates for PND. They surveyed more than 3 000 physicians across Canada, finding marked variation in their views on the value and effectiveness of various prenatal diagnostic techniques; on the criteria for referring pregnant women to genetics centres; on the nature and severity of disabilities; and on termination of severely affected pregnancies. authors note that, overall, physicians in Canada have a cautious attitude toward PND. They view it as a serious undertaking and do not take referrals to genetics centres lightly. They find that, while views differ according to specialty, the regional differences are so striking that they are moved to talk about "provincial cultures" in the use of PND. particularly significant that the variation in regional views that the authors note coincides with the regional variation in referrals noted by Dr. Hamerton and colleagues. This is of concern, as decisions connected with PND should be made by the woman involved, not by her physician, as this is an infringement of her personal autonomy. Taken together, the data raise questions about the similarity of treatment in terms of obtaining referral and access to prenatal diagnostic services a pregnant woman over the age of 34 would receive across the country.

Prenatal ultrasound can be used as a diagnostic tool in the case of women already known to be at higher risk of a congenital anomaly or genetic disease, but it can also be used as a screening device to help identify otherwise unsuspected conditions. The value of ultrasound in diagnosis is acknowledged, but there is debate about its value as a routine screening procedure for all pregnant women. Geoffrey Anderson analyzed data on the use of ultrasound for this purpose in Ontario and British Columbia during the 1980s; his findings demonstrate that current practice with regard to the use of prenatal ultrasound for screening is unacceptable in terms of quality of care and effective use of scarce health care resources.

Dr. Anderson found that utilization rates in both provinces doubled over the period under study, but while expenditures on ultrasound doubled in British Columbia, they quadrupled in Ontario. He attributes this finding to the increased provision of ultrasound outside hospital settings, which is permissible in Ontario. Also importantly, Dr. Anderson finds that the doubling in the use of ultrasound has taken place in the absence of conclusive evidence of the value of such tests to the health of the mother or the fetus.

Dr. Anderson notes that the use of ultrasound has increased to the point where there is almost a *de facto* screening policy being followed by practitioners and funded by provincial health ministries, but this policy is being applied unevenly. Some women have as many as four ultrasounds during their pregnancy, while others do not have any; some have ultrasound early in their pregnancy, while many do not have it until later.

He concludes that there are fundamental decisions facing physicians and health ministries about the use of prenatal ultrasound as a screening procedure in Canada. If it is decided that the benefits of ultrasound screening for maternal and fetal health are marginal and that there is no justification for using ultrasound in this way, there should be a dramatic reduction in the overall level of use. On the other hand, if it is decided that such a screening program is justified and required, the distribution of ultrasound should change, so that all women are offered at least one ultrasound, fewer have multiple ultrasounds, and more women have early, as opposed to late, ultrasounds.

Dr. Anderson's findings point to the need for the same careful evaluation of prenatal screening technologies as has been applied to prenatal diagnostic technologies. Bernard Chodirker and Jane Evans provide an example of such evaluation — in this case, of the MSAFP screening program offered in Manitoba. MSAFP screening, which permits early detection of chromosomal syndromes and neural tube defects in the fetus, is part of routinely offered prenatal care in many parts of Europe and North America. Manitoba has the only provincial MSAFP screening program in Canada. Their analysis of that province's experience permits Drs. Chodirker and Evans to set out some of the conditions that must be met before such screening can be offered on a population basis in a coordinated, timely, and ethical fashion.

The authors note a clear impact of the screening program in terms of a reduced prevalence of neural tube defects at birth and a potential impact in terms of Down syndrome. They also point to some problems with the program, in particular with regard to informed consent. Disturbingly, they find that fewer than 40 percent of physicians seek patients' specific consent for testing, and that more than two in five do the test automatically unless the patient specifically declines. They point to the need both for increased physician education on the procedure and for increased information and counselling for women undergoing the screening — considerations that are consistent with experiences relating to the use of other prenatal testing procedures.

Conclusion

Taken together, the studies in this volume document how PND is currently practised in this country. The data raise some important issues that should be addressed.

The first concerns equality of access to PND. The likelihood of referral for PND differs markedly according to the region of the country in which a woman resides. Differences in the attitudes of referring physicians are likely to be more of an obstacle than differing desires for access on the part of pregnant women or lack of services. The evidence is clear that ultrasound is being offered on an inappropriate basis, even if it is considered of value as a routine screening procedure. Some women are

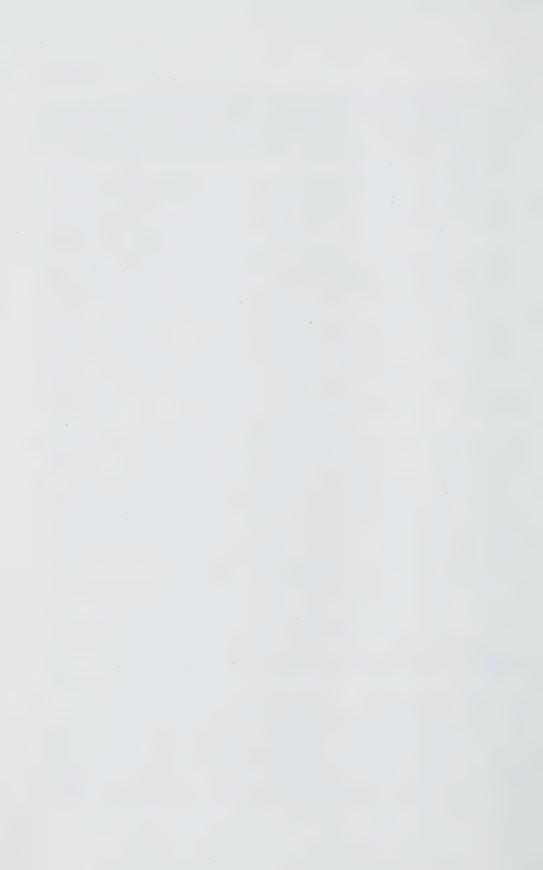
receiving multiple ultrasounds, and most are receiving at least one, but a significant minority are receiving none. Given that more than \$100 million each year is spent on ultrasound, in spite of the absence of any conclusive evidence as to its effectiveness as a screening tool, it is an issue that should be addressed.

Advances in screening programs such as MSAFP testing raise the possibility that abnormal results may be a more effective criterion for referral for PND than simply the criterion of advanced maternal age. But, as the studies on ultrasound and MSAFP testing indicate, there is a need to ensure that prenatal screening programs receive the same rigorous evaluation before wider dissemination that prenatal diagnostic procedures performed at genetics centres have done. This is, of course, more difficult to control when the physicians involved are dispersed in the community.

Another issue concerns the context in which testing, whether screening or diagnostic, is offered. The studies of PND in Canada and of Manitoba's MSAFP screening program indicate wide variations in the information provided to women considering PND or undergoing screening. It is disturbing that the informational material provided to assist patients in making informed choices is often too complex and technical to be easily understood. It is also disturbing that, outside genetics centres, physicians are not seeking specific consent to MSAFP testing.

It is important that the large number of physicians who refer patients for PND or who offer ultrasound or MSAFP screening to their patients be educated and be made aware of the need to respect women's autonomy. It is evident that more needs to be done to ensure that physicians are aware of what tests are relevant to pregnant women, which pregnant women should be offered them, and what kinds of information should be made available. The correspondence by region between the differences in referral rates for PND and the personal views of physicians on disability or abortion is troubling. It is important that it be the choice of the woman, not the physician, to pursue or not pursue PND.

The studies in this volume paint a picture of services that are valued by many women and are used with care and without coercion at the genetics centres, but whose provision outside the centres is of some concern. They point to the need to remain vigilant and to continue to subject PND techniques to rigorous evaluation with regard to both their effectiveness and their social impact.





Prenatal Diagnosis in Canada — 1990: A Review of Genetics Centres

John L. Hamerton, Jane A. Evans, and Leonie Stranc



Executive Summary

Prenatal diagnosis for genetic disease is now a routinely offered part of antenatal care for women in Canada who are at increased risk. In 1990, at least 22 222 women were referred for prenatal diagnostic services through genetics centres in Canada. This is about 5 percent of all pregnant women. Of these women, 78 percent were referred because of their advanced maternal age (AMA). The remaining 22 percent were referred for a variety of reasons, including a previous chromosome abnormality (2.4%), familial chromosome abnormality (2.3%), abnormal maternal serum alpha-fetoprotein (AFP) (3.6%), abnormal ultrasound (3.1%), single gene disorders (1.6%), and possible teratogen exposure (1.4%).

Twenty-two sites offering prenatal diagnostic services could be found in all regions of Canada except Prince Edward Island, New Brunswick, the Northwest Territories, and the Yukon. Amniocentesis and cytogenetic analysis were available at all of these sites, while chorionic villus sampling (CVS) was available at only 11. There were 35 formal outreach sites associated with genetics centres. The most extensive network was in Alberta, which had 18 outreach sites,

compared to 8 in Ontario, 4 in the Maritimes, 3 in Newfoundland, and 6 in British Columbia.

Sixty-four laboratories performed genetic testing in Canada in 1990; 25 were cytogenetic laboratories, 10 were molecular, and 12 were biochemical, while 17 performed maternal serum AFP testing. In Canada as a whole, 52 percent of women eligible for prenatal diagnosis because of advanced maternal age were referred for testing. This figure ranged from a high of 64 percent in Quebec to lows of 15 percent in Newfoundland and 22 percent in Saskatchewan.

Access to prenatal testing for a particular woman has been shown to be determined by several factors, including her knowledge of and desire for testing, the availability of health care providers who have an adequate knowledge of testing so that an appropriate referral can be made, her physician's awareness of testing and willingness to refer, personal biases concerning testing and its possible outcomes, and the perception of the risks involved. These factors clearly interact to determine whether a woman is tested or not, and indicate that there is a need for education about prenatal testing at all levels.

Despite the generally high quality of service provided in Canada, some improvements should be made: access to these services was not equal across the country, particularly in Newfoundland, the Maritime provinces, and Saskatchewan, where only 1.5 to 3 percent of pregnant women are referred, compared to about 6 percent in the other regions. More research is needed to determine the reasons why women in these three areas are less often referred for prenatal diagnosis than women in the rest of Canada. Once this is determined, steps need to be taken by their provincial health care systems to rectify the situation.

As techniques improve and appropriate pilot studies are done, non-invasive testing, such as ultrasound scanning, maternal serum AFP screening, and triple testing (maternal serum AFP, beta human gonadotropin, and estriol) for Down syndrome and other common chromosome anomalies, will become increasingly important. Maternal serum AFP screening is now available in Manitoba and parts of Ontario and takes place on an ad hoc basis in other parts of Canada. There is little doubt in our minds that many women who are screened do not receive adequate counselling or follow-up. It is our view that all parties involved should review the guidelines established by the American Society of Human Genetics, which have also been affirmed by the Canadian College of Medical Geneticists (CCMG), and either adhere to these guidelines or, if that is not possible, confine the use of maternal serum AFP determination to women at high risk for neural tube defects.

With the advent of relatively routine use of ultrasound screening during pregnancy it is inevitable that it may become a tool for the detection of fetal anomalies; it is already used on occasion to inappropriately reassure patients they do not have a fetus with Down syndrome. Before these practices become more widespread, the CCMG and the Society of Obstetricians and Gynaecologists of Canada (SOGC) should develop guidelines for the use of ultrasound in the detection of fetal anomalies.

The next step in the development of non-invasive screening tests is triple testing. Appropriate pilot studies should be undertaken before the introduction of such screening programs, and when such programs are introduced they should be integrated with the genetic services in the area to ensure appropriate counselling and follow-up.

This study demonstrates the variability in both quality and quantity of the data collected by the centres. It is our view that the continued development of prenatal services should be monitored regularly. For this to be done effectively, centres should standardize data collection across the country. The CCMG Prenatal Diagnosis Committee and the SOGC Genetics Committee should determine what the minimum data requirements are for such monitoring, and provide centres with standardized data collection forms. A national registry should be established.

In general, prenatal testing is being used in Canada in a responsible, conscientious, and sensitive manner. Only one centre indicated that it strongly discouraged testing in the absence of willingness to terminate an affected pregnancy. The centres are aware of the potential for misuse, especially in respect to sex selection, and exercise strong control over the availability of testing for non-medical reasons. Only one instance of invasive prenatal testing for sex selection was documented in Canada in 1990. Two major legal or ethical issues were identified by centres — sex selection and the issue of confidentiality.

Prenatal diagnosis for genetic disorders is a routinely offered part of antenatal care for many women and, as new tests are introduced, it will become a medically indicated procedure for an increasing number of pregnant women. To meet the needs of this increased load on an already overburdened service, staffing will have to be supplemented. We suggest that genetic counsellors should play a greater role, dealing with most referrals and follow-up, with physician geneticists handling cases requiring more complex diagnostic assessment.

Physician and public education is urgently needed to ensure that prenatal tests are understood by all involved, and that women desiring testing receive appropriate referral. So that programs develop in an integrated and cost-effective fashion, it would seem vital that communication be improved between medical geneticists, community health care providers, pregnant women and their partners, and provincial and federal health care agencies. Only if there is a conscious attempt to make improvements will these programs continue to develop to meet the medical, social, and emotional needs of all Canadian women.

Recommendations

General

1. To ensure adequate development of prenatal diagnostic services for genetic disease to continue to meet the medical, social, and emotional

needs of Canadians in the future, appropriate committees should be established at both the federal and provincial levels to evaluate the services provided on an ongoing basis and to recommend and monitor the introduction and spread of new technologies in this field. Such committees should have representation from the medical genetics community, including physicians, laboratory directors, scientists, researchers, and genetic counsellors in addition to obstetricians, family practitioners, public health workers, health care economists, ethical and religious advisors, and members of the lay public.

- 2. A code of practice governing provision of services for genetic prenatal diagnosis should be drawn up with consultation from the CCMG, the SOGC, and the Canadian College of Family Physicians (CCFP) and should be adhered to by individuals and centres offering such services. Individuals or programs that cannot, for whatever reason, follow such a code of practice should refer their patients to other health care providers when a recognized medical indication exists.
- 3. Given the variability of the data bases maintained in different centres and the different degrees of difficulty in quickly obtaining such data, the CCMG Prenatal Diagnosis Committee should be requested to identify a list of variables that should be obtained from all prenatal diagnosis patients, and a generalized format that can be used to collect and store these data. If a standardized method of data collection is used by all centres, it should prove possible to develop a national registry for prenatal diagnosis patients and monitor outcomes.

Accessibility

- 1. To ensure equality of access to prenatal programs for all women, centres should establish outreach programs so that counselling and informational packages can be made available to rural women close to their homes. These should be written in an accessible way and not require high levels of education to understand them. Coordination to avoid expensive duplication of effort by centres in this regard is essential.
- 2. Provincial advisory committees should be developed or reinstated to coordinate the provision of genetic services and eliminate unnecessary duplication.
- 3. Canadian medical schools should be encouraged to include among continuing medical education offerings courses on all aspects of the delivery and application of prenatal testing, including ethical implications.
- 4. Women's groups and others concerned about the health and well-being of women should be encouraged to develop objective and

balanced informational modules about prenatal testing, including maternal serum AFP screening, which can be made available to women through their physicians' offices, public health units, antenatal classes, and other means appropriate to each particular region of Canada.

- 5. The provincial colleges of physicians and surgeons, and medical associations should emphasize to their members that failure to discuss with their patients the option for referral for a medically indicated prenatal diagnostic service is unethical and constitutes bad medical practice.
- 6. Evidence from the Atlantic provinces suggests that lack of availability of obstetricians or family physicians in particular health districts may result in lower than expected utilization of prenatal services in these regions. If this finding can be shown to apply to the rest of Canada, then centres, in conjunction with their provincial health services, should try to ensure that at least someone in each public health or other appropriate health unit is knowledgeable concerning prenatal diagnosis and the options open to women at risk, so that women wishing such testing can obtain counselling near home and be referred to an appropriate centre for testing.
- 7. Centres with large immigrant populations in their catchment areas should ensure that written material and, in particular, consent forms are available in the most frequently used languages.

Invasive Prenatal Diagnostic Procedures

- Given that CVS has been shown to be a relatively safe and reliable technique for first trimester prenatal diagnosis, centres should make this technique more widely available to women, especially those in high-risk groups, such as those who have previously had an affected child or who are at risk of having a child with a single gene disorder.
- CVS should be restricted to major centres where the number of tests
 performed will ensure that the obstetricians performing the test have
 sufficient experience. Centres offering this procedure should limit the
 number of operators to ensure that each has sufficient experience with
 the testing procedure.
- 3. Where CVS is medically indicated and not available in the local centre, provincial health care plans should meet both the cost of travelling to the nearest centre where the test is available and the out-of-province testing costs.
- 4. Given the provincial responsibility for the delivery of health care services, we recommend that interprovincial barriers be removed to allow each woman to receive prenatal testing in the most appropriate

- centre dealing with her particular problem. Accessibility would therefore not be denied if a local centre were unable to perform the testing.
- 5. Because the period between testing and the provision of results is a time of heightened anxiety for the woman, all centres need to be concerned about the length of time taken to provide results. It is our view that results from a second trimester amniocentesis for advanced maternal age should take no longer than three weeks, and results from CVS for the same indication, no longer than two weeks.
- 6. Laboratory techniques used for prenatal diagnosis and the protocols used to evaluate unusual results, including mosaicism, are variable. The Cytogenetics Committee of the CCMG is in the process of establishing guidelines in this area as to the best way to approach this evaluation. All centres should be encouraged to apprise themselves of this information and follow recommended protocols when they have been finalized.

Non-Invasive Testing

- 1. The CCMG and SOGC should evaluate the practice of using ultrasound screening to rule out chromosome abnormalities, in particular Down syndrome, and should prepare guidelines to ensure that such ultrasound screening is applied appropriately.
- 2. Given the different protocols used to offer maternal serum AFP screening across the country, the test should be offered only on a population basis, within the confines of a program that adheres to the guidelines established by the American Society of Human Genetics (American Society of Human Genetics 1987; Garver 1989) and affirmed by the CCMG (Davidson 1987). Where the resources to develop such programs and the associated counselling are not available, the test should be restricted to patients at high risk.

Pregnancy Outcomes

- 1. All centres should declare as explicit policy that agreement to terminate a pregnancy is not a precondition or requirement for undergoing prenatal testing.
- 2. All Canadian women, wherever their location, should have reasonable access to prenatal testing and be aware of the options open to them after learning the test results. Those opting for termination of pregnancy should be provided with the necessary referral to achieve that option and should not normally have to travel out of province to obtain the service.

- 3. Where they do not already exist, all centres providing prenatal testing should have, within their centre or by referral, facilities to provide women and their partners with both pre- and post-termination counselling, including grief counselling.
- 4. Given the relatively high frequency of chromosomal anomalies other than Down syndrome and trisomy 18 detected by invasive testing, all women considering prenatal diagnosis should be informed initially of the possibility of such diagnoses and that, in some cases, parental karyotyping may be required before definitive counselling is possible.
- 5. In view of the possibility that there is an increased risk of limb deficiency defects after CVS, particularly when performed very early in gestation, we recommend that the procedure not be performed before 10 weeks' gestation until definitive epidemiological information is available.

Decision-Making Process

1. It is our view that all centres should formalize an appropriate committee structure to ensure that prenatal policy is regularly and adequately discussed. This committee should include not only caregivers, but also someone with some knowledge of ethical principles and representatives of consumer and women's groups. If it is not thought appropriate to include consumer groups on the committee, arrangements should be made at least annually to meet with such groups to learn of their concerns.

Staffing

- 1. Adequate fellowship training support should be provided by the federal and provincial governments to provide training for accreditation by the CCMG for Ph.D.s wanting to work in genetic service laboratories.
- 2. Universities should consider development of suitable interdisciplinary training programs at the master's level for individuals wishing to undertake careers as genetic counsellors.
- 3. The CCMG and the Canadian Association of Genetic Counsellors (CAGC) should be encouraged to include genetic counsellor training programs in the CCMG accreditation of centres program.
- 4. The CAGC should be encouraged to develop an accreditation program for Canadian genetic counsellors, and each centre should be encouraged to develop an appropriate career structure for such individuals, if not already done, either through an affiliated university or hospital or through their provincial program, whichever is the most appropriate.

5. Genetics centres should be encouraged to review their programs to ensure that appropriately trained staff are filling positions and, when hiring new personnel, that the most cost-effective solution is followed.

Introduction

Prenatal diagnostic services have been available to Canadian women since about 1973, when the Medical Research Council of Canada commissioned the first Canadian trial to assess the safety and accuracy of amniocentesis as a means of obtaining fetal tissue to diagnose chromosome disorders (Medical Research Council of Canada 1977; Simpson et al. 1976). The first Canadian guidelines for the delivery of prenatal diagnostic services were published in 1974 (Hamerton et al. 1974) and were a joint effort of the Genetics Society of Canada, the Canadian Paediatric Society, and the Society of Obstetricians and Gynaecologists of Canada (SOGC). This was the first attempt in the world to establish national guidelines for service delivery in this area. These guidelines were updated in 1983 and again in 1991 (CCMG and SOGC 1991; SOGC 1983).

Canada recently completed the first randomized clinical trial of chorionic villus sampling (CVS) compared to second trimester amniocentesis (Canadian Collaborative CVS-Amniocentesis Clinical Trial Group 1989; Lippman et al. 1992). At the present time, a proposal for a clinical trial comparing early amniocentesis with second trimester amniocentesis is under consideration by the Medical Research Council of Canada (R.D. Wilson, pers. comm., 1991).

Thus, Canada has been a leader in the field of safety and clinical testing of prenatal diagnostic techniques before their introduction to clinical practice. In 1987, the Canadian College of Medical Geneticists (CCMG) did a survey of prenatal diagnostic centres in Canada to determine what services were being delivered. However, a comprehensive survey of the current state of prenatal diagnostic services, their accessibility to all Canadian women, their cost, the nature of the counselling offered, reasons for referral, and future needs has never been adequately conducted.

The mandate of the Royal Commission on New Reproductive Technologies was to undertake a broad and comprehensive study of the new reproductive technologies, including the prenatal diagnosis of genetic disease, from a scientific, social, and legal perspective, and to make recommendations to the Government of Canada concerning the development and control of these technologies in Canada. The present study will provide a comprehensive data base on Canadian prenatal diagnostic services that will allow the Commission to examine the current state of this service in Canada and make appropriate recommendations for the future.

Background and Literature Review

Few detailed studies on national prenatal diagnostic services have been done. The Royal College of Physicians of London established a working group in the United Kingdom to review service delivery and to make recommendations (Royal College of Physicians of London 1989). This group identified the objectives of prenatal diagnosis as (i) allowing the widest possible range of informed choice to women and couples at risk of having children with an abnormality; (ii) providing reassurance and reduction of anxiety level associated with reproduction; (iii) allowing couples at risk to embark on a family knowing they may avoid the birth of seriously affected children through selective abortion; and (iv) ensuring optimal treatment of affected infants through early diagnosis.

These objectives are by and large acceptable to the Canadian genetics community, which has recently summarized the Canadian position in a revised set of guidelines for the delivery of these services (CCMG and SOGC 1991) and produced a statement of principles aimed at both physicians and the lay public (CCMG 1991). The recommendations contained in the report of the Royal College working group included recommendations relating to equality of access, the development of a policy advisory structure, the development of a code of practice, provision of resources, development of genetic counselling services, and the development of a team approach to the provision of services in the health regions (Royal College of Physicians of London 1989).

Various groups appearing before the Commission during public hearings expressed concern about prenatal diagnostic services as they are presently delivered in Canada, and occasionally revealed a significant level of misunderstanding concerning both the service and its overall objectives (CCMG 1991). During these hearings the Commission heard statements indicating a perception that some centres required women to agree to undergo a termination of pregnancy if an abnormality was detected before allowing the testing. Concern about the extent of sex selection for non-medical reasons was also raised on several occasions.

Another issue raised during the hearings was the possible devaluation of human life as a direct result of our ability to detect abnormalities prenatally and terminate affected pregnancies. The National Action Committee on the Status of Women (National Action Committee on the Status of Women 1991) and other women's groups called for a moratorium on any further developments in prenatal testing pending development of appropriate guidelines or legislation.

Ethical and potential eugenic issues raised by prenatal diagnosis have been recently discussed on several occasions (Beck 1990; Clarke 1990; Smith and Miller 1990). Beck likened genetic amniocentesis, followed by termination of pregnancy when an abnormal fetus was detected, to the negative eugenic policies followed in many countries between 1900 and

1950, which reached extremes in Nazi Germany between 1930 and 1945. In response, the CCMG issued a reasoned statement concerning the objectives, nature, and scope of prenatal diagnostic services in Canada (CCMG 1991).

In 1989, Roy and Hall reviewed medical genetics services in Canada and, by means of a questionnaire, sought the opinions of Canadian medical geneticists on several ethical dilemmas as part of an international crosscultural study of ethics and medical genetics (Roy and Hall 1989; Wertz and Fletcher 1989). Two issues of immediate relevance to this study were, first, whether an agreement to terminate a pregnancy where an affected fetus was detected should be a requirement before agreeing to prenatal diagnosis, and, second, the issue of sex selection for non-medical reasons. On the first issue, an overwhelming proportion of Canadian medical geneticist respondents indicated that prior agreement to termination should not be a condition of performing the test. On the second, about 30 percent of respondents indicated that they would perform prenatal diagnosis when the sole objective was to select the sex of the baby, and a further 17 percent indicated that they would refer. Such opinions were usually based on the grounds of patient autonomy. Fifty-three percent indicated that they would refuse to perform testing on these grounds.

Canada has a universal system of health care; medical genetics forms part of that health care system (Science Council of Canada 1991), which, although funded in part by the federal government by means of transfer payments, is administered by each of the 10 provincial governments and the two territorial governments. In theory, universality and maintenance of standards are ensured by the Canada Health Act. The current fiscal problems faced by our health system mean that each service must be looked at not only in terms of social justice and benefit, but also in terms of cost and the cost-benefit accruing to the population. Because of the existence of this universal health care system, Canadian medical geneticists have also been concerned about the standards of delivery of genetic services to the Canadian population, and the training of Canadian medical geneticists. In 1975, the CCMG was established to accredit centres for both training and service. It has also been responsible for the establishment of guidelines for service delivery and, when necessary, the establishment of codes of practice for genetic services (Miller 1979).

The CCMG was incorporated with the following objectives (Miller 1979):

- "1. defining the characteristics of medical genetic centres and the responsibility of these to health care;
- 2. accrediting medical genetic centres providing health care services;
- 3. informing appropriate levels of government of the role of medical geneticists in health care;

- informing appropriate levels of government on the nature and extent of medical genetic services that should be provided for each 4. province:
- when requested, aiding in negotiations with appropriate levels of 5. government on methods of funding; and
- issuing certificates of accreditation in medical genetics to M.D.s 6. and Ph.D.s who possess the necessary qualifications."

In the context of the present report, items 2 and 6 above are important. Both M.D.s and Ph.D.s are examined individually in several sub-disciplines of medical genetics, including clinical genetics, medical genetics, cytogenetics, biochemical genetics, and molecular genetics. Training programs are available in nine centres (American Society of Human Genetics 1990). People undertaking such a program and achieving a passing grade in the examinations are awarded the Fellowship of the Canadian College of Medical Geneticists, implying that they have achieved a certain standard of knowledge in their specialty. In 1989, the Royal College of Physicians and Surgeons of Canada, at the urging of the CCMG, established a specialty in medical genetics open to M.D.s who undertake the appropriate training and pass the Royal College examinations.

The CCMG, through its Accreditation of Centres Committee, also undertakes the evaluation of centres for the delivery of service. Accreditation involves the completion of a detailed self-study followed by a site visit. Centres satisfying the accreditation team are accredited for five years. Accreditation is voluntary, and centres may lose accreditation in a particular sub-specialty if they do not have a CCMG-qualified staff person in that specialty. When the committee has concerns and believes that problems it has identified can be rectified within a specific period, it may award provisional accreditation pending correction. With the exception of Memorial University of Newfoundland, Dalhousie University, Laval University, the University of Montreal, and the University of Saskatchewan, all of the university centres are accredited. None of the general hospital centres is accredited and none has applied for accreditation. It must be stressed that because a centre is not accredited, it does not necessarily indicate a lowering of the standard of service provided to the public.

In Canada, as elsewhere, the accuracy and safety of prenatal testing have been of concern: in 1973 the Medical Research Council of Canada established a small working group to determine the safety and accuracy of genetic amniocentesis. The group reported in 1975 that amniocentesis, although carrying a small risk of miscarriage generally put at between a half and one percent above random risk, was overall a safe and accurate prenatal test for chromosome abnormalities (Medical Research Council of Canada 1977; Simpson et al. 1976). Studies in the United Kingdom, the United States, and Denmark reached similar conclusions (NICHD 1976; Tabor et al. 1986: Medical Research Council 1978).

In 1984, when chorionic villus sampling was introduced into clinical practice, concerns were also expressed about its safety. The first randomized clinical trial comparing chorionic villus sampling to genetic amniocentesis was designed in Canada and made possible by a voluntary agreement between all Canadian centres that such a procedure would be available only within the context of the trial. The results of this trial indicated that, although CVS carried a slightly higher (but not significant) risk of miscarriage than genetic amniocentesis, and its level of accuracy was perhaps slightly lower, it was sufficiently safe and accurate to make it an acceptable alternative for first trimester diagnosis (Canadian Collaborative CVS-Amniocentesis Clinical Trial Group 1989; Lippman et al. 1992; "Chorion Villus Sampling" 1991).

A randomized trial in the United Kingdom reached similar conclusions, although its risk estimates were considerably higher and did show a significant difference from genetic amniocentesis (Medical Research Council 1991). The need for an early prenatal test is accepted and, with improvements in the resolution of ultrasound, early amniocentesis is a possibility (Jorgensen et al. 1992; Thayer et al. 1990). This test has been introduced in many international centres, especially in the United States. Canada is planning a clinical trial to determine the safety and accuracy of this procedure before it is introduced into routine practice here (R.D. Wilson, pers. comm., 1991). For now, Canadian guidelines indicate that amniocentesis should be performed for routine indications between 15 and 17 weeks' gestation.

Maternal serum alpha-fetoprotein (AFP) screening for neural tube defects, introduced into clinical practice in the late 1970s (Wald et al. 1977b) in many parts of North America and Europe, is now a routine part of prenatal care (American College of Obstetricians and Gynecologists 1986). By the mid-1980s it had become clear that maternal serum AFP screening also provided information about the risk of carrying a fetus with a chromosome abnormality (Cuckle et al. 1984, 1987; Hershey et al. 1986; New England Regional Genetics Group 1989), the risk of fetal demise, and some other abnormal pregnancy outcomes (Brock et al. 1977; Katz et al. 1990; Lidbjörk et al. 1977; Macri et al. 1978; Wald et al. 1977a; Waller et al. 1991). Thus, this non-invasive prenatal testing procedure, if appropriately interpreted, can provide both information concerning specific abnormalities and more general information concerning fetal well-being.

Such information might assist in pregnancy management if women whose levels are outside the norm receive proper counselling and follow-up. Wilson (1992) surveyed 19 Canadian centres offering maternal serum AFP testing through a questionnaire. In 1989, 50 180 women received maternal serum AFP screening in eight provinces. In Manitoba, the only province with a province-wide screening program, 9 300 women were screened in 1989; this represented about 60 percent of the births in that province. It was clear from this study that the extent of maternal serum AFP screening varied between provinces, and only in Manitoba and Ontario had any

attempt been made to organize testing in an integrated manner and on a province-wide basis. The Manitoba experience has been reviewed by Chodirker and Evans (1993) for the Commission.

The present study was designed to conduct a detailed assessment of the current state of Canadian prenatal diagnostic services so that the Commission might have a comprehensive data base reflecting current practice and policies on which useful recommendations for the future might be based. A summary of the results of the study is presented below.

The recommendations presented in this report represent the opinions of the authors. They are based on an integration of the results of this investigation with the authors' previous knowledge of Canadian genetic prenatal diagnostic services.

Methods

A workshop was held at the Commission offices on 29-30 April 1991 with all centre directors or their representatives in attendance. Details of the study were explained at that time and amendments to protocol agreed upon. The major change in protocol was to attempt to collect data from the 1990 calendar year rather than 1989 as originally proposed, to ensure a more up-to-date data base. The disadvantage was that the outcome data were likely to be less complete. However, it was believed to be important that the survey include the most up-to-date data possible, even at the expense of less complete outcomes.

The first two months were used to design the questionnaires for the site visit report and the collection of statistical data, to begin construction of two dBase IV data bases to house this information, and to organize the site visit schedule. The site visit protocol was piloted in June, using the Winnipeg centre as a test for the practicality of the approach to be used.

All 22 Canadian genetics centres offering prenatal diagnostic services agreed to participate in the study. Sixteen of the centres were situated in the Canadian universities having medical schools, and six were in large community hospitals. Data collected from each centre refer to the period from 1 January to 31 December 1990. Each centre to be surveyed was visited by the study coordinator, with most visits lasting a minimum of two days. Interviews were held with the prenatal diagnosis coordinator, the centre director, relevant laboratory directors, and other individuals as appropriate to the particular centre administration. Upon completion of a site visit, the study coordinator compiled a site visit report (Appendix 1), which was then forwarded to the centre director for review, clarification or correction of errors, and the addition of any comments that the director thought might be required. The data obtained from each centre and any comments received were entered into a dBase IV data base for further analysis. In addition to the site visits, statistical data relating to the 1990

calendar year were collected by a mail-in questionnaire (Appendix 2). These were distributed to centres in July 1991 with a requested return date of 31 October 1991. Most questionnaires were returned by 31 December 1991, although one centre did not return its information until March 1992. On receipt, all data were entered into a separate dBase IV data base for analysis. Statistical summaries were done using the SPSS statistical analysis package for desktop computers (1990 Statistical Package for the Social Sciences). One centre was visited a second time to extract data from charts, since it did not have the staff to provide the data required.

The data obtained from each centre varied in quality and completeness, some centres having more comprehensive data bases than others. In 1990, few centres maintained extensive computerized data bases and the amount and type of information collected varied considerably between centres. The data presented in this report reflect the quality of those data.

Results

Genetics Centres in Canada Providing Prenatal Diagnosis Services

Twenty-two sites in Canada where prenatal diagnostic testing was done in association with a genetics centre in 1990 were identified (Figure 1, Table 1). Of the 22 centres, 10 were accredited by the CCMG for the delivery of service (Appendix 3). Sixteen were university medical centres or tertiary care hospitals associated with university medical centres. Six were large community hospitals, sometimes also associated with universities, sometimes not. No centre doing prenatal testing existed in Prince Edward Island, New Brunswick, Labrador, the Northwest Territories, or the Yukon. Although women from Prince Edward Island and New Brunswick were referred to Halifax for testing, amniocentesis was available to women in all provinces (see Table 2). Women from the Northwest Territories were usually referred to Edmonton or Winnipeg, depending on the region of the territories from which they originated. Some amniocenteses were also performed in Yellowknife and the fluids sent to Edmonton for analysis. Women from the Yukon were tested in Vancouver. Referrals from Labrador were handled in St. John's, Newfoundland.

The availability of CVS was limited (Table 3); many centres had limits on the number of procedures offered. CVS was not available in Saskatchewan, Newfoundland, or Calgary (except for high-risk cases). The Calgary centre has been conducting a trial of early amniocentesis between nine and 13 weeks' gestation to replace CVS (Iwanicki et al. 1992). Only one province, Manitoba, had a provincial maternal serum AFP screening program, funded from a separate budget by the provincial health plan. Maternal serum AFP screening was also performed in some parts of

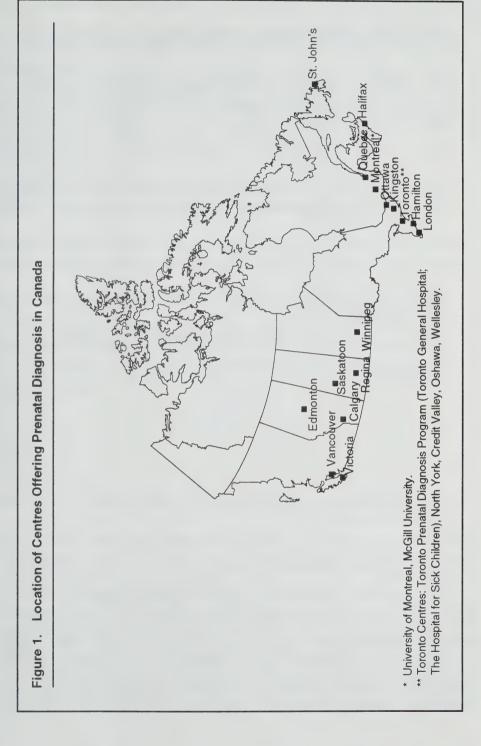


Table 1. Major Centres Offering Prenatal Diagnostic Services in 1990

				Ontar
British Columbia	Alberta	Saskatchewan	Manitoba	Toronto
Vancouver Victoria	Calgary Edmonton	Saskatoon Regina	Winnipeg	Toronto Prenatal Diagnosis Prograr Wellesley Credit Valley North York Oshawa

^{*} The Toronto Prenatal Diagnosis Program includes both Toronto General Hospital and The Hospital for Sick Children.

Table 2. Sites Remote from a Major Genetics Centre Where Amniocenteses Were Performed in 1990

Alberta	Northwest Territories	Ontario
Fort McMurray Grande Prairie	Yellowknife	Sudbury Thunder Bay Sault Ste. Marie

Ontario, particularly Toronto and London, but no integrated provincial program existed.

Most provincial centres had some form of outreach program delivering genetic and prenatal services to remote areas (Table 4). Outreach programs were best developed in Ontario, Alberta, and British Columbia. The type of service provided varied widely from outreach nurses in public health units who would handle routine referrals (Ontario, Alberta, British Columbia) to centres with no on-site support but regular visits from a geneticist. The proportion of prenatal patients seen on these visits was usually small, since the timing of prenatal counselling and testing falls into

^{*} Not an exhaustive list.

cluding Toronto	— Quebec	Maritimes	Newfoundland
ngston ndon milton tawa	Univ. of Montreal McGill Laval	Halifax, NS	St. John's

uebec*	Maritimes	Newfoundland
imouski hicoutimi aie-Comeau ouyn-Noranda ept-Îles es de la Madeleine t-Hyacinthe ngava Bay affin Island herbrooke	Moncton, NB Saint John, NB	Corner Brook

a relatively narrow time frame. If prenatal testing was not available locally, patients would travel to the nearest centre.

In Newfoundland, the Maritimes, and Quebec, prenatal outreach primarily consisted of accepting fluids from outside the main centre for analysis. Outreach in Manitoba and Saskatchewan was not well developed. In Manitoba, with the exception of a few amniocenteses done through obstetric outreach, women travelled to Winnipeg for counselling and invasive testing. In Saskatchewan, an independent cytogenetics laboratory in Regina processed local amniotic fluid samples, but most counselling was done by local obstetricians. Sudbury and Victoria also had cytogenetic

Table 3. Availability of Chorionic Villus Sampling, by Region, in 1990

			Ont	ario	
British Columbia	Alberta	Manitoba	Toronto	Excluding Toronto	Quebec
Vancouver	Edmonton Calgary*	Winnipeg	Toronto Prenatal Diagnosis Program**	Kingston London Hamilton Ottawa	Univ. of Montreal McGill Laval

* Reduced availability; the test was not performed for routine advanced maternal age.

** The Toronto Prenatal Diagnosis Program encompasses a joint program offered through The Hospital for Sick Children and Toronto General Hospital.

laboratories with minimal on-site support. Both sites were in transition in 1990; Sudbury was becoming a referral centre for Northern Ontario with its own cytogenetics laboratory and counselling support, and Victoria had lost its geneticist and cytogeneticist, becoming an outreach site.

Funding for outreach centres came from a variety of sources: it was included in the global funding for genetics in St. John's and Saskatoon. Both Quebec and Alberta had provincial genetic networks that funded outreach and general genetic programs. In British Columbia, outreach was funded as a special project of the Ministry of Health; in Ontario, funding was given directly to the public health units. Ontario's outreach began as a joint venture between the Association of Genetic Counsellors of Ontario (AGCO) and the Imperial Order of the Daughters of the Empire (IODE). The IODE provided start-up funding for the network, which was then taken on by the Ministry of Health. The interest of the IODE in genetic services in Ontario over the years has been of enormous value.

All of the centres with formal outreach programs reported some form of provincial advisory committee, with many reporting directly to the Minister of Health. In both Ontario and Alberta, two of the provinces with the largest and best organized outreach services, these committees were recently disbanded at the request of the provincial governments.

A summary of the major characteristics of each genetics centre offering prenatal testing is given in Appendix 3.

Table 4. Formal Outreach Sites Associated with Genetics Centres

Alberta	British Columbia Alberta Ontario	.o.	Maritimes	Newfoundland
Mountainview	Sudbury	ıry	Saint John, NB*	Corner Brook*
Lethbridge Medicine Hat	Thunde	Thunder Bay	Charlottetown, PEI*	Grand Falls*
Red Deer	Timmins	IIS	Moncton, NB*	
Drumheller	North Bay	Bay		
Sturgeon Health Unit	Orillia			
Athabasca Health Unit	Windsor	or		
Peace River Health Unit		Peterborough		
South Peace Health Unit	.=)		
Fort McMurray				
Alberta West Central Health	ealth			
Unit				
High Level - Fort Vermilion	lion			
Leduc - Strathcona Health	alth			
Unit				
Stony Plain - Lac Ste. Anne	Inne			
Northeastern Alberta				
Wetoka Health Unit				
Alberta East Central Health	alth			
Unit				
Minburn - Vermilion Health	alth			
Unit				

This unit had no outreach nurse but was visited regularly by a geneticist.

Recommendations:

To ensure equality of access to prenatal programs for all women, centres should establish outreach programs so that counselling and informational packages can be made available to rural women close to their homes. These should be written in an accessible way and not require high levels of education to understand them. Coordination to avoid expensive duplication of effort by centres in this regard is essential.

Provincial advisory committees should be developed or reinstated to coordinate the provision of genetic services and eliminate unnecessary duplication.

Laboratories

Cytogenetics

In 1990, 64 laboratories were involved in prenatal diagnostic testing in Canada (Table 5). Twenty-five laboratories performed cytogenetic testing, all of which provided cytogenetic analysis of amniotic fluid samples. Fifteen laboratories performed analysis of chorionic villus samples. This reflects both the more limited availability of CVS across the country and the fact that the analysis of such samples is more labour-intensive than analysis of amniotic fluid samples. Significant experience with the culturing of samples obtained at early amniocentesis (9-13 weeks) was available in Calgary where a local assessment of this alternative early test was under way (Iwanicki et al. 1992).

Molecular Genetics

There were 10 laboratories performing molecular diagnoses for a variety of genetic diseases (Appendix 4). With the exception of Newfoundland, Manitoba,* and Saskatchewan, at least one molecular diagnostic laboratory existed in each province. In Ontario, there were four molecular diagnostic laboratories. These four laboratories formed a provincial network, each undertaking the diagnosis of specific genetic diseases for the province as a whole: Kingston dealt with disorders on the long arm of the X chromosome, Ottawa performed prenatal diagnosis for myotonic dystrophy, Hamilton performed prenatal diagnosis for haemoglobinopathies, and The Hospital for Sick Children provided facilities for cystic fibrosis, Duchenne muscular dystrophy, and disorders on the short arm of the X chromosome.

^{*} Manitoba established a Molecular Diagnostic Laboratory in 1992.

Table 5. Reporting of Laboratories Involved in Prenatal Testing in Canada (N)

	Pathology/ laboratory medicine	Genetics	Genetics/ pathology/ laboratory medicine	Other	Total
Cytogenetics	12	7	6	0	25
Molecular genetics	4	4	2	0	10
Biochemical genetics	3	4	3	2*	12
Maternal serum AFP	12	2	2	1**	17
Total	31	.17	13	3	64

* Children's Psychiatric Research Institute, London, Ontario; Atlantic Research Centre, Halifax, Nova Scotia.

** Cadham Provincial Public Health Laboratory, Winnipeg, Manitoba.

Three laboratories for molecular testing existed in Quebec as part of the Quebec Network of Genetic Medicine. Again, regional specialization existed for deoxyribonucleic acid (DNA) diagnostics in this province, with cystic fibrosis testing available at McGill, myotonic dystrophy at Laval, and Duchenne muscular dystrophy at the University of Montreal (some samples were also sent to Toronto in 1990). The other three molecular laboratories were located in Halifax, Calgary, and Vancouver. However, access to molecular diagnosis was not as limited as this distribution might suggest, since these laboratories usually accepted samples for analysis from outside their own province.

Fifteen centres routinely banked DNA from families with genetic diseases that are or may shortly become diagnosable by molecular means (Appendix 2). Ten of the 15 centres had molecular laboratories. The six centres that did not bank DNA included three that primarily provided a cytogenetic service (Wellesley, Regina, Victoria), one small genetics centre (Saskatoon), and the Toronto General and Credit Valley Hospitals, which referred molecular diagnoses to The Hospital for Sick Children.

Recognizing the importance of this resource to future generations, the CCMG recently published guidelines for DNA banking and molecular genetic diagnoses (Hall et al. 1991) outlining the standards that should be maintained by these services, and gave sample consent forms to be used by centres with DNA-banking operations.

Biochemical Genetics

Twelve laboratories were performing biochemical testing for a variety of inborn errors of metabolism in 1990 (Appendix 5). Two laboratories were located in Halifax, three in Quebec, four in Ontario, and one each in Winnipeg, Saskatoon, and Vancouver. Limited availability for testing existed in Calgary. Most centres not having laboratories of their own referred samples for testing to a laboratory of their choice. Access to these tests for women at risk of having a child with a biochemical genetic disease was therefore widely available in Canada, despite limitation in the number of laboratories. It can be argued that, given the rarity of some of these conditions, increasing the number of laboratories would neither be cost-effective nor improve the quality of service, since the number of tests done by any given laboratory would then be so small as to be inefficient and very costly.

Maternal Serum Alpha-Fetoprotein

Maternal serum AFP screening on a province-wide basis was available only in Manitoba. In Toronto and some parts of southern Ontario, many women were screened but there was no organized and integrated screening program. In most other provinces, maternal serum AFP testing was mainly offered to women at high risk for neural tube defects. In most if not all provinces (with the exception of Manitoba), limited use was made of the information provided by maternal serum AFP, including the use of low values for predicting an increased risk of chromosome abnormalities. All provinces had laboratories testing levels of both maternal serum AFP and amniotic fluid AFP. The Manitoba screening program was coordinated through Genetics and was a multidisciplinary program, with the screening being done by the Cadham Provincial Public Health Laboratory and follow-up of abnormal pregnancies by the Fetal Assessment Unit (Chodirker and Evans 1993).

Laboratory Organization

The organization of the 64 genetic diagnostic laboratories varied between centres (Table 5). Of the 25 cytogenetics laboratories, 12 reported through the departments of pathology or laboratory medicine, and had only informal relationships with the genetics centres; 7 laboratories were integral components of the respective genetics centre; and 6 had a dual relationship with the genetics centre and departments of pathology or laboratory medicine. Of the 25 cytogenetics laboratories, 21 had directors accredited by the CCMG in the specialty of cytogenetics. Two small laboratories did not have a professionally trained cytogeneticist as director and reported through hospital departments of pathology or laboratory medicine.

There were 10 laboratories providing a molecular diagnostic service. All provinces except Newfoundland, Manitoba, and Saskatchewan had at least one such diagnostic laboratory. Several of the larger molecular laboratories accepted significant numbers of referrals from outside provincial boundaries. Seven of the 10 laboratories had CCMG-accredited directors.

Molecular diagnosis is a new and emerging but extremely powerful technology, which will come to play an ever increasing role in genetic diagnosis. Because of the excellent referral mechanisms in Canada, access to these services was probably adequate for most women. Because of the cost of establishing these laboratories, and the relative rarity of individual conditions amenable to diagnosis by molecular means, it may be most appropriate to establish a series of large regional laboratories, each serving a supra-provincial area. However, as our health care services are a provincial responsibility, major changes would be required to funding and billing arrangements and agreements among the provinces to support such laboratories jointly, if this mode of organization were to be effective.

Twelve laboratories provided prenatal diagnosis for inborn errors of metabolism using biochemical methods. Five directors were accredited by the CCMG; the remainder were Ph.D. biochemists. Six laboratories were part of the departments of Laboratory Medicine or Clinical Chemistry although three others also reported to Genetics. In Halifax, one laboratory was part of the Atlantic Research Centre, while a second was housed in the children's hospital and reported to the department of laboratory services. At the University of Montreal, McGill, Laval, and The Hospital for Sick Children in Toronto, the laboratory was part of the genetics section. In London, Ontario, the laboratory was part of the Children's Psychiatric Research Institute. All of these laboratories accepted out-of-province referrals.

Seventeen laboratories assayed maternal serum and amniotic fluid AFP: 12 were part of departments of pathology or laboratory medicine; 2 were an integral part of their respective genetics centre; and in one province, Manitoba, which had a provincial screening program, the assays were done in the Cadham Provincial Public Health Laboratory (Chodirker and Evans 1993). Laboratory directors were, in general, Ph.D. biochemists. Interpretation of the results of maternal serum AFP assays varied between centres. In some, the results were forwarded to the physician of record; in others, a more detailed interpretation was provided (ibid.). In Ontario, private laboratories with no affiliation with the genetics centres were also offering maternal serum AFP screening, but we have no information on their staff or protocols.

Referral of Women for Prenatal Diagnosis

Source of Referrals

As far as we have been able to determine, at least 22 222 women were referred for prenatal diagnostic services through genetics centres in Canada in 1990. The sources of referral for these patients included general medical practitioners, obstetricians, other physicians, and specialized medical services such as fetal assessment units and screening programs. A small proportion of women were self-referred. Where available, records of the

referring source indicated different patterns of referral in different parts of

the country.

Tables 6 and 7 indicate the number of women referred by source and centre and the proportions referred by certain groups. Over 56 percent of patients with known referral sources were sent by obstetricians. This varied from a high of over 84 percent in North York, Ottawa, and Saskatoon to less than 25 percent in British Columbia. Referrals from general practitioners also varied widely from apparently negligible numbers in Edmonton to nearly 80 percent of prenatal diagnosis patients seen at Grace Hospital in Vancouver, with a national figure of 40 percent. These data may be partly influenced by differences in record keeping. It is likely that the role of the general practitioner and obstetrician in referral of patients for prenatal diagnostic services varies according to local health care practices. In Edmonton, for example, general practitioners apparently refer such patients first to obstetricians who then make appropriate arrangements for counselling and testing, but in Calgary, general practitioners refer directly to genetic services.

In many centres most patients were referred directly by general practitioners, obstetricians, or both; in others, such as Edmonton, a relatively high proportion were referred from other sources. Most referrals were still primarily from the medical community from other physicians, maternal serum AFP screening programs, or fetal assessment units. Few patients were directly referred by public health nurses or through outreach programs. Although the questionnaires sought information on referrals from community clinics and women's health clinics, the centres did not report any direct referrals from these sources. Also, the number of women reported as self-referred was less than 1 percent of the total. Again, it is likely that the coding of referral data by centres is masking the true referral patterns in certain cases. For example, in the Winnipeg centre the physician responsible for direct patient care and to whom results are to be forwarded is noted as being either a general practitioner or obstetric specialist: the individual or agency that generated the request for prenatal diagnostic services is not recorded. Similarly, only in a few cases were we able to determine whether a referral for services was precipitated by a woman herself or occurred after a discussion initiated by her physician or other health care advisor. In addition, no information could be obtained on the proportion of women eligible for prenatal diagnostic services who declined referral for counselling or testing.

Demographic Parameters

Most centres kept some demographic information on their prenatal diagnosis patients in a readily accessible form. Thus, data are available on the ages of referred patients in 92 percent of cases (Table 8) and on their previous reproductive history in over 50 percent of cases (Tables 9 and 10). In 1990, about two-thirds of all referrals were women between 35 and 39

Table 6. Number of Women Referred, by Source and Centre (% = no. referred from source/total of known referral sources)

	Herer ger pract	нетеггед by general practitioner	Refer	Referred by obstetrician	All	All other sources	Total no. of known	No. with multiple	Referral	
Centre	z	%	z	%	z	%	sources	sources	source	of women
Vancouver	385	64.8	146	24.6	63	10.6	594			594
Grace	1 477	79.5	382	20.5			1 859		9	1 865
Victoria									243	243
Edmonton			352	76.9	106	23.1	458		518	926
Calgary	591	72.2	213	26.0	15	1.8	819			819
Saskatoon	15	8.8	154	90.6	_	9.0	170			170
Regina									134	134
Winnipeg	322	32.9	929	67.1			826	10		968
London	313	38.6	454	56.0	44	5.4	811			811
Hamilton									930	930
Ottawa	77	7.8	882	89.2	30	3.0	686	40	259	1 606
Kingston	69	41.8	96	58.2			165			165
Toronto General					295*		295		2 195	2 490

Table 6. (cont'd) (% = no. referred from source/total of known referral sources)

	Referred by general practition	Referred by general practitioner	Refer obste	Referred by obstetrician	Alle	All other sources	Total no. of known	No. with multiple	Referral	Total no.
Centre	z	%	z	%	z	%	sonices	sources	unknown	of women
Sick Children's	13	26.5	18	36.7	18	36.7	49	-	127	175
Wellesley	09	20.1	235	78.9	က	1.0	298			298
Oshawa	422	52.7	285	35.6	94	11.7	801	332		469
Credit Valley	550	53.3	475	46.1	9	9.0	1 031		402	1 433
North York	167	9.6	1 626	84.2	139	7.2	1 932	135		1 797
U. of Montreal	086	38.4	1 563	61.3	7	0.3	2 550		200	2 750
McGill									2 026	2 026
Laval									729	729
Halifax	232	38.7	366	61.0	0	0.3	009		52	652
St. John's	31	22.5	103	74.6	4	2.9	138	16		122
Total	5 704		8 006		827	6	14 537	534	8 219	22 222

Referred for abnormal maternal serum AFP.

Total 63 90 614 30 94 1 128 Misc. (public health nurses, outreach, 9 30 45 Self-referred က 22 313 353 fetal assessment Referred by units 9 6 69 Table 7. Breakdown of Other Referrals, by Centre (N) through maternal serum AFP Referred 295 77 135 507 by "other" physicians Referred 100 22 ന 154 Diagnosis Program Toronto Prenatal U. of Montreal Credit Valley North York Saskatoon Vancouver Edmonton St. John's Wellesley Oshawa Calgary Centre London Halifax Ottawa Total

Table 8. Age of Women Referred, by Region (% = no. referred for that group/total over all age groups)

	British Columbia	Alberta	Saskatchewan	Manitoba
No. of women aged under 15 years at delivery (%)				1 (0.1)
No. of women aged 15-19 at delivery (%)	7 (0.3)	6 (0.5)	5 (2.9)	15 (1.5)
No. of women aged 20-24 at delivery (%)	18 (0.7)	73 (5.7)	10 (5.9)	36 (3.7)
No. of women aged 25-29 at delivery (%)	68 (2.3)	106 (8.3)	21 (12.4)	76 (7.9)
No. of women aged 30-34 at delivery (%)	125 (5.1)	173 (13.6)	21 (12.4)	176 (18.2)
No. of women aged 35-39 at delivery (%)	1 854 (75.4)	786 (61.7)	100 (58.8)	561 (58.0)
No. of women aged 40-44 at delivery (%)	372 (15.1)	126 (9.9)	13 (7.6)	99 (10.2)
No. of women aged over 44 years at delivery (%)	9 (0.4)	4 (0.3)		4 (0.4)
Total no. age known	2 453	1 274	170	968
Age unknown	249	521	134	
Total no. of women	2 702	1 795	304	968

^{*} McGill over-reported by nine.

	Ontario	-			
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
3	2	3	not known		9
17 (0.3)	5 (0.1)	43 (0.8)	not known	2 (1.6)	100
134 (2.2)	50 (1.4)	213 (3.9)	not known	15 (12.3)	549
332 (5.4)	172 (4.9)	515 (9.4)	not known	17 (13.9)	1 307
629 (10.2)	262 (7.5)	790 (14.5)	not known	17 (13.9)	2 193
4 193 (67.9)	2 415 (68.8)	3 330 (61.1)	348	58 (47.5)	13 645
847 (13.7)	577 (16.4)	545 (10.0)	59	12 (9.8)	2 650
19 (0.3)	27 (0.8)	14 (0.3)	3	1 (0.8)	81
6 174	3 510	5 453	410	122	20 534
488	2	61	242		1 697
6 662	3 512	5 514*	652	122	22 231*

Table 9. Gravid Status of Referred Women, by Region

(% = no. referred for that group/total over all reported gravid states)

	British Columb	ia Alberta	Saskatchewan	Manitoba
No. first pregnancy (%)		188 (21.7)		97 (14.9)
No. second pregnancy (%)		251 (29.0)		197 (30.3)
No. third pregnancy (%)		202 (23.3)		182 (28.0)
No. fourth pregnancy or more (%)		226 (26.1)		174 (26.8)
Unknown	2 702	928	304	318
Total	2 702	1 795	304	968

Table 10. Parity of Referred Women, by Region

(% = no. referred for that group/total of all reported parities)

	British Columb	ia Alberta	Saskatchewan	Manitoba
Parity = 0 (%)		372 (34.0)		161 (25.2)
Parity = 1 (%)		392 (35.8)		259 (40.6)
Parity = 2 (%)		216 (19.7)		150 (23.5)
Parity ≥ 3 (%)		114 (10.4)		68 (10.7)
Unknown	2 702	330	304	330
Total	2 702	1 795	304	968

Ontario					
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
584	466	732	107	25	2 199
(19.9)	(18.3)	(24.6)	(19.6)	(27.2)	
853	754	875	161	30	3 121
(29.1)	(29.7)	(29.5)	(29.4)	(32.6)	
732	612	680	127	18	2 553
(25.0)	(24.1)	(22.9)	(23.2)	(19.6)	
762	710	683	152	19	2 726
(26.0)	(27.9)	(23.0)	(27.8)	(20.6)	
3 731	970	2 535	105	30	11 623
6 662	3 512	5 505	652	122	22 222

Ontario					
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
987	848	1 086	273	38	3 765
(43.8)	(32.8)	(36.6)	(41.9)	(41.3)	
696	931	1 070	208	25	3 581
(30.9)	(36.1)	(36.0)	(31.9)	(27.2)	
432	525	559	102	19	2 003
(19.1)	(20.3)	(18.8)	(15.6)	(20.7)	
141	278	255	69	10	935
(6.3)	(10.8)	(8.6)	(10.6)	(10.9)	
4 406	930	2 535	0	30	11 938
6 662	3 512	5 505	652	122	22 222

years of age and a further 13 percent were 40 years of age or older. The proportion of "older" women was highest in British Columbia and Ontario and lower in Newfoundland, Quebec, and Manitoba. Saskatchewan and Alberta appeared to have relatively high numbers of younger referrals. However, in these two centres data on age were not available for 29 percent and 45 percent of women, respectively. Manitoba had the highest proportion of referrals of women 30 to 34 years of age, which was probably a result of the existence of the maternal serum AFP screening program and thus reflected the referral of women being found at increased risk of having a fetus with Down syndrome.

Although data are incomplete on prior reproductive histories in referred women, the distribution of gravid status and parity in cases where these parameters are known was relatively similar across the country (Tables 9 and 10). The proportion of referred women documented as being in their first pregnancy was 20.7 percent nation-wide, ranging from a low of 15 percent in Manitoba to 27 percent in Newfoundland. Figures were even more consistent for second (mean 29.4%, range 29% to 33%), third (mean 24.1%, range 20% to 28%), and fourth or more (mean 25.7%, range 21% to 28%) pregnancy status. Thus, prenatal diagnosis referrals were usually made for women who had had at least one prior pregnancy, and half of the referred women had had two or more pregnancies.

Assuming that the women in whom gravid status (Table 9) is known are also those for whom parity data were provided (Table 10), it would appear that a relatively high proportion of women referred for prenatal diagnosis had previous reproductive losses. Nation-wide, 36.6 percent apparently had not had a successful pregnancy. Comparing this figure with the 20.7 percent referred in their first pregnancy would indicate either a high degree of fetal loss or preferential recording of pregnancy histories that indicate reproductive misfortune. Both of these factors were presumably acting.

There was less discrepancy between the proportions of women referred in their second or third pregnancy and those referred after one or two previous deliveries. However, the number of referred women with three or more previous births (9.1%) was considerably lower than the number reported as being in their fourth or later pregnancy (25.7%). It is more difficult to postulate recording bias alone as an explanation for this discrepancy and, given the relatively high frequency of spontaneous abortion (approximately 15%) and its increasing frequency with maternal age, it is likely that many women referred with four or more pregnancies had suffered at least one prior fetal loss.

Many of the reasons women are referred for prenatal diagnosis infer a high potential for previous reproductive loss (e.g., previous chromosomal abnormality, structural chromosomal rearrangements in family members, previous history of neural tube defects) but it is not yet possible to analyze in detail the relationship between demographic factors including age, gravid status and parity, and reason for referral. What can be stated is that most

women referred for prenatal diagnosis in 1990 had experienced previous pregnancies and, in several cases, at least one pregnancy loss.

Reasons for Referral: Advanced Maternal Age

Reasons for referral are known for over 91 percent of patients (Table 11). As anticipated, the most common reason for referral was advanced maternal age (AMA), which accounted for 77.7 percent of referrals in total, ranging from 87.5 percent in British Columbia to 53.3 percent in Newfoundland. There was a tendency for regions that had a high number of referrals to have a higher proportion of AMA cases.

All centres offered amniocentesis and almost all had a cut-off for referral for amniocentesis for late maternal age of 35 years of age at the estimated time of delivery. For a short period in 1990, the McGill centre reduced the cut-off to 34.2 years, while in Victoria it was 35 years at the time of amniocentesis. In both centres it is now 35 years of age at the time of delivery.

There was more variation in the cut-offs used for chorionic villus sampling. In most centres offering the test, the cut-off was 35 years. However, several centres restricted this procedure to somewhat older women: age 37 years at Toronto General Hospital, 39 years at the University of Montreal and Laval, and 40 years at McGill. Laval noted that it had previously raised the cut-off for chorionic villus sampling from age 37 years, and Toronto General Hospital has also raised the cut-off from age 37 to 39 years since mid-1991. All centres but two reported that maternal age alone was the standard parameter used to determine risks for Down syndrome. The North York and Manitoba centres also used maternal serum AFP values in combination with maternal age to determine risk in some cases. No centre reported other biochemical markers in clinical service, although some may have had research protocols for triple testing.

Proportions of Eligible Women Referred for Prenatal Diagnosis

It was not possible to estimate the exact proportion of eligible (i.e., 35 years of age at estimated time of delivery) women referred for prenatal testing in 1990 because detailed birth statistics for 1990 are not yet available for many jurisdictions; many prenatal diagnosis patients seen in 1990 would not have delivered until 1991; and prenatal referrals in early pregnancy cannot be directly related to births in any one year. However, some estimation of these data can be obtained by comparing numbers and proportions of live births at different maternal ages with prenatal diagnosis referrals. Table 12 shows live births by age of mother for 1989, the last year for which data are currently available from Statistics Canada.

In Canada in 1989, 32 240 (8.2%) of births were to women 35 years of age or older. Assuming no change in birth rate or age-specific fertility for 1990 and ignoring potential prenatal losses due to spontaneous or induced abortion and stillbirth, a similar number of women in these age groups might have been expected to be pregnant and eligible for prenatal diagnosis

Table 11. Reasons for Referral of Women for Prenatal Testing, by Region (% = no. referred for an indication/total of known indications)

Number of women referred for	British Columbia	Alberta	Saskatchewan	Manitoba
Advanced maternal age (%)	2 358 (87.5)	987 (71.5)	206 (68.4)	658 (66.9)
Previous chromosome abnormality (%)	77 (2.9)	28 (2.0)	12 (4.0)	18 (1.8)
Parental chromosome abnormality (%)	30 (1.1)	6 (0.4)	(0.3)	6 (0.6)
Relative with chromosome abnormality (%)	33 (1.2)	36 (2.6)	2 (0.7)	12 (1.2)
Abnormal maternal serum AFP (%)	26 (1.0)	9 (0.7)	6 (2.0)	113 (11.5)
Previous/family history of neural tube defects (%)	49 (1.8)	10 (0.7)	16 (5.3)	40 (4.1)
Inborn error of metabolism (%)	4 (0.1)	10 (0.7)	6 (2.0)	2 (0.2)
Other single gene disorder (%)	10 (0.4)	25 (1.8)	1 (0.3)	14 (1.4)
Disorder with chromosome marker/abnormality (%)	1 (0.0)	2 (0.1)		
Maternal/paternal irradiation (%)	2 (0.1)	17 (1.2)	2 (0.7)	(0.1)
Abnormal ultrasound (%)	71 (2.6)	20 (1.4)	24 (8.0)	57 (5.8)
Teratogen exposure (%)	8 (0.3)	143 (10.4)	4 (1.3)	12 (1.2)
Sex for medical reasons (%)	5 (0.2)	14 (1.0)		1 (0.1)
Other indications (%)	20 (0.7)	73 (5.3)	21 (7.0)	50 (5.1)
No. multiply referred	378	103	8	16
Reason unknown	386	518	11	
Total of known reasons	2 694	1 380	301	984

Table 12. Live Births, by Age of Mother and Region

(% = no. of births for that group/total over all age groups)

	British Columbia	Alberta	Saskatchewan	Manitoba	a Ontario
Births to mothers under 15 (%)	23 (0.1)	35 (0.1)	26 (0.2)	· 30 (0.2)	39 (0.0)
Births to mothers 15-19 (%)	2 535	3 153	1 684	1 686	7 228
	(5.8)	(7.3)	(10.1)	(9.7)	(5.0)
Births to mothers 20-24 (%)	9 167 (20.9)	9 725 (22.4)	4 377 (26.3)	4 150 (24.0)	28 283 (19.5)
Births to mothers 25-29 (%)	16 054	16 222	6 332	6 196	57 003
	(36.7)	(37.4)	(38.0)	(35.8)	(39.2)
Births to mothers 30-34 (%)	11 664	10 938	3 341	3 924	38 825
	(26.6)	(25.2)	(20.1)	(22.7)	(26.7)
Births to mothers 35-39 (%)	3 840	2 947	794	1 177	12 270
	(8.8)	(6.8)	(4.8)	(6.8)	(8.4)
Births to mothers 40-44 (%)	465	316	92	156	1 628
	(1.1)	(0.7)	(0.6)	(0.9)	(1.1)
Births to mothers over 44 (%)	14 (0.0)	15 (0.0)	5 (0.0)	1 (0.0)	41 (0.0)
Age of mother unknown (%)	7 (0.0)			1 (0.0)	21 (0.0)
Births to mothers 35 and over (%)	4 319	3 278	891	1 334	13 939
	(9.9)	(7.6)	(5.4)	(7.7)	(9.6)
Total	43 769	43 351	16 651	17 321	145 338

Source: Statistics Canada, 1989 data.

in 1990. As far as can be determined, approximately 16 803 such referrals were made, or about 52 percent.

Comparing data on live births and referrals across the country indicated several interesting findings. The variation in the proportion of patients referred because of late maternal age seemed to reflect real differences in the proportion of older women giving birth. Thus, provinces such as British Columbia and Ontario with the highest proportions of births to older women (9.9% and 9.6%, respectively) had the highest proportion of referrals for late maternal age (87.5% and 78.5%, respectively). Similarly, provinces with a lower proportion of births to older

Quebec	Maritimes	Northwest Territories	Yukon	Newfoundland	Total
30	22	7	2 (0.4)	12	226
(0.0)	(0.1)	(0.5)		(0.2)	(0.1)
3 841	2 040	279	37	913	23 396
(4.2)	(8.5)	(18.9)	(7.7)	(11.8)	(6.0)
20 445	6 380	440	114	2 305	85 386
(22.1)	(26.4)	(29.7)	(23.8)	(29.7)	(21.7)
39 162	9 170	444	152	2 683	153 418
(42.4)	(38.0)	(30.0)	(31.7)	(34.6)	(39.1)
22 393	5 088	221	130	1 415	97 939
(24.2)	(21.1)	(14.9)	(27.1)	(18.2)	(24.9)
5 751	1 247	73	41	393	28 533
(6.2)	(5.2)	(4.9)	(8.5)	(5.1)	(7.3)
713 (0.8)	181 (0.8)	12 (0.8)	(0.8)	38 (0.5)	3 605 (1.0)
17 (0.0)	3 (0.0)	1 (0.1)		3 (0.0)	100 (0.0)
21 (0.0)	6 (0.0)	2 (0.1)	′ .		58 (0.0)
6 481 (7.0)	1 431	86	45	434	32 238
	(5.9)	(5.8)	(9.3)	(5.6)	(8.2)
92 373	24 137	1 479	480	7 762	392 661

mothers, such as the Maritimes (5.9%), Newfoundland (5.6%), and Saskatchewan (5.4%), had lower percentages of patients referred because of late maternal age (68.5%, 53.3%, and 68.4%, respectively). In two provinces, this pattern was not obviously maintained. In Manitoba, the proportion of births to older mothers was intermediate (7.7%); however, the proportion of prenatal diagnosis referrals that relate to late maternal age appeared relatively low (66.9%). The finding is somewhat artefactual because of the high number of referrals for abnormal maternal serum AFP in this province (11.5%). Advanced maternal age cases comprised 75.5 percent of non-maternal serum AFP-related referrals in Manitoba. In

Quebec, on the other hand, the proportion of births to older mothers was less than average (7%) but the proportion of referrals for late maternal age

was relatively high (76.9%).

Although the number of 1989 live births to older mothers and 1990 referrals for late maternal age can only be crudely equated, these data are likely proportionately similar and their comparison provides interesting information about potential referral for prenatal diagnosis related to late maternal age in Canada. The highest ratio of such 1989 births to 1990 referrals (64.5%) is seen in Quebec, suggesting a high utilization of prenatal diagnostic services for women eligible on the basis of age in this province. Ratios of 49 percent to 57 percent are seen for British Columbia, Manitoba, and Ontario. The ratios are approximately 30 percent for the Maritimes and Alberta, 23 percent for Saskatchewan, and 15 percent for Newfoundland. Simplistically, these figures appear to suggest underutilization of prenatal diagnostic services by older women in some areas. However, Newfoundland and Saskatchewan were also provinces with proportionately fewer births to older women, suggesting a general trend that the fewer births there are to older women, the less likely such women are to be referred for prenatal counselling. However, a survey of this type cannot explore in depth the reasons for apparent lower utilization of services in some provinces and higher utilization in others. As only known referrals are documented, different proportions of women in different regions may be informed of the availability of testing or may decline to be referred for further evaluation, or both.

If the numbers of all women referred for prenatal diagnosis in 1990 are compared with total 1989 births, it is apparent that the provincial trends are not confined to referrals for late maternal age. Quebec, Ontario, Manitoba, and British Columbia all have ratios suggesting referral of approximately 6 percent or more of pregnant patients, Alberta 4 percent, the Maritimes 3 percent, Saskatchewan 2 percent, and Newfoundland 1.5 to 2 percent.

Other Reasons for Referral for Prenatal Diagnosis

Table 11 documents in detail numbers of referrals for other reasons in addition to AMA. For situations such as previous chromosomal abnormality, chromosomal abnormality in a parent or other relative, single gene disorders, parental irradiation, and sex determination for medical reasons, the proportion of referrals was similar across the country. However, in other categories there are differences in referral patterns, presumably reflecting local areas of interest and expertise, different incidences of high-risk situations, and variable availability of specialized services.

Maternal serum AFP screening was available as a provincial program in Manitoba and to many pregnant women in Ontario, especially in the Toronto area. Thus it was not unexpected that a relatively high proportion of referrals in these regions was for evaluation of abnormal AFP levels. In

Calgary a high proportion of referrals was for teratogen exposure, perhaps reflecting local interest or awareness. Few of these women subsequently had invasive diagnostic tests. Also, there were major differences in the proportion of women referred because of abnormal ultrasound findings (e.g., over 10% in the Maritimes and Newfoundland but only 0.1% in Alberta).

There may be many reasons behind these differences. For example, in some areas the practice might have been to refer all such cases directly to genetics centres for further evaluation; in others, the local obstetricians or general practitioners might have taken more direct responsibility for patient management. Use of ultrasonographic examination may have varied from being a relatively routine part of prenatal care — leading to its use, deliberate or otherwise, as a screening test for abnormalities of fetal morphology and growth or abnormal amniotic fluid volume, which often led to further investigation — to being infrequent due to limited availability of the technology.

One final category, family history of neural tube defect, also showed interprovincial differences. The proportion of such referrals was high in Quebec, Newfoundland, and the Maritimes, where the incidence of this type of birth defect was relatively high. It was also relatively high in Saskatchewan. Perhaps the known increased recurrence risk and the availability of a highly sensitive prenatal diagnostic test for this disorder led to a higher proportion of eligible women with this indication being referred in Saskatchewan compared to other lower-risk situations such as late maternal age. Manitoba also had a relatively high proportion of referrals for neural tube defect, which may have been due partly to greater awareness of the availability of prenatal diagnosis for this condition by physicians using maternal serum AFP screening in their practices.

Nation-wide, 6.6 percent of referred women were known to have more than one indication for prenatal diagnostic evaluation. This figure was especially high in British Columbia (16%) and Newfoundland (15%). Much of the variability in centres' data with respect to this question probably relates more to differences in record keeping than to major differences in the complexity of cases referred for prenatal diagnosis. However, several centres did indicate that certain groups of patients were over-represented in their catchment/service area due to a high incidence of certain genetic disorders or a large population of a specific ethnic group at increased risk

for a genetic disease.

For example, The Hospital for Sick Children, North York, Toronto General Hospital, Wellesley, Winnipeg, and Vancouver, all centres with relatively high immigrant populations or clientele, commented on the need for thalassaemia screening for Asian populations; the Winnipeg centre, The Hospital for Sick Children, and the McGill centre also mentioned Mediterranean populations in this regard. Winnipeg, The Hospital for Sick Children, and McGill also mentioned Tay-Sachs screening for Ashkenazi Jewish populations. Other populations considered at risk for specific

disorders included Mennonites, with Tourette's syndrome and diabetes more commonly found in this group in Calgary, hypophosphatasia in Manitoba and Saskatchewan Mennonites, and cystic fibrosis and galactosaemia in the Mennonite populations served by Hamilton and London in Ontario. Hutterite populations in western Canada are also at risk for Bowen-Conradi disease and cystic fibrosis. Native Canadians were perceived to be at increased risk in certain centres but the reasons varied between regions. Sikhs were noted to be at increased risk for neural tube defects by the Vancouver and Credit Valley centres.

Non-Medical Reasons for Requests for Testing

All centres reported that they used the indications given by Canadian guidelines for prenatal diagnosis to screen referrals for invasive procedures and attempted to discourage referral of women who did not meet the guidelines. This screening was often done by prenatal diagnosis coordinators or office staff when speaking by telephone with the physician's office or the patient. The calls were not usually documented so it is impossible to determine the number of such requests that were refused. However, most centres were prepared to see and counsel such patients if the family or physician was insistent.

The two most common indications for referral for non-medical reasons listed by centres were maternal anxiety and sex selection. At least 282 patients were seen in genetics centres for maternal anxiety. In most centres, staff would counsel the family about the relative risks of amniocentesis and fetal anomaly. Several centres would offer maternal serum AFP screening to determine if the patient was at increased risk of having a child with Down syndrome or would offer ultrasound scanning to provide reassurance. One centre (Calgary) offered triple testing; 164 amniocenteses were performed because this test indicated an increased risk for Down syndrome. Some centres noted that an amniocentesis may have been offered if the patient was 33 or 34 years of age, or had a family history of mental retardation or physical handicap.

Other centres noted that an amniocentesis was offered occasionally if the laboratory was not overloaded or, if the patient was prepared to pay, the sample would be taken and sent to a private laboratory. In British Columbia, at least one obstetrician was performing amniocenteses for non-medical indications and sending fluid samples to the United States. Other centres noted that they would provide the telephone numbers for centres in the United States to patients upon request. In Manitoba, only 10 patients were seen for maternal anxiety, all of whom were tested; in Saskatchewan, only two patients were seen and both were tested. In Alberta, 19 of 26 referred patients were tested. In Ontario, centres outside Toronto saw at least 75 patients and apparently all were tested, while in Toronto, where the 35-year guideline was applied quite strictly in some centres, at least 12 of the 37 patients seen were refused testing and, in Quebec, although 133 patients with maternal anxiety were seen, only 25

percent were tested. Only two chorionic villus sampling tests were offered for anxiety, one in Quebec and one in Ontario.

All centres reported that referral for sex selection for non-medical reasons was a rare occurrence and strongly discouraged. Many centres reported no such referrals in 1990, although tentative requests by telephone may have been received and rejected without documentation. These patients were rarely counselled, although some centres would provide the telephone numbers of U.S. prenatal programs if asked. Vancouver's centre also noted that some physicians in the United States were offering Canadians sex selection information using ultrasound. Throughout the country, we were able to document 14 women counselled in 1990 whose reason for requesting testing was sex selection: six in Quebec, five in British Columbia, and three in Toronto. In all but one case invasive testing was refused. As details of the case tested could potentially lead to a breach in medical confidentiality, they will not be provided. However, the woman was considering termination of pregnancy for social reasons. The test indicated the fetus was not of the desired sex, but no information is available on the pregnancy outcome.

Changes in Referral Patterns

Most centres kept ongoing local statistics on prenatal patients and could assess objectively changes in the number and type of prenatal diagnosis patients referred over time. Others could make more subjective evaluations.

All centres except the one in Saskatoon reported a significant increase in demand for prenatal diagnostic services in the five years leading up to 1990. Saskatoon also noted a steady but apparently not significant increase in amniocenteses from 111 in 1985-86 to 160 in 1990-91. Eleven centres reported an increase in referrals for AMA, six mentioned molecular diagnosis, and two referred to assessment of metabolic/biochemical problems becoming more common. One centre noted that the increasing use of ultrasound and maternal serum AFP had made amniocenteses for family history of neural tube defects less common. At least two centres noted they were seeing more patients referred for abnormal ultrasound findings and one mentioned low maternal serum AFP values leading to increased demand.

Many centres noted that no major changes in the referral patterns had been observed for five years. However, eight centres reported that more physicians were referring patients. Five specifically commented that more general practitioners were sending patients for counselling, while in Saskatoon more obstetricians were referring. One centre noted more self-referrals.

Some centres commented that an improved awareness of prenatal diagnosis indications among physicians influenced the demand for prenatal diagnosis, and one noted that the addition of a physician of a particular ethnic background to their staff led to an increase in patients belonging to

that group. Most centres reported that they made an effort to educate health care professionals in their area about new developments in prenatal diagnosis including rounds and seminars, newsletters, and articles in local medical bulletins. Increased patient awareness was also mentioned and, at the McGill centre, the presence of a teratogen enquiry hotline for pregnant women led to an increase in referrals for drug exposure during pregnancy.

Direct education of the public by genetics centres is not common; however, patients with clinically significant family histories who were being followed up by the genetics clinics often were encouraged to contact the clinic if they became pregnant or were contemplating a pregnancy to see if new tests were available. Occasionally, centres contacted specific families when prenatal diagnosis became available: neurofibromatosis and fragile X syndrome were noted by Credit Valley in this regard, and McGill informed families with cystic fibrosis patients when testing became an option.

Almost all centres thought that the demand for prenatal diagnostic services was likely to continue to increase over the next five years. In some cases, it was noted that demand for testing based on AMA might start to level off but that requests for molecular testing were likely to increase, especially if there were further breakthroughs in research. An increased demand for chorionic villus sampling was also anticipated. Saskatoon noted that it was likely to see a higher proportion of eligible patients but, since its population was declining, no major increase in demand was expected.

Accessibility

MacLeod et al. (1993) have examined the geographic pattern of utilization and referral rates for prenatal diagnostic services in 1990 using data collected during this survey on the postal codes of clients referred for prenatal diagnosis. To determine the number and distribution of physicians likely to refer, a data base listing every family physician, general practitioner, obstetrician, and gynaecologist was purchased. At the time of writing, analysis has been completed for the Atlantic provinces only.

Using ecological modelling, MacLeod and colleagues have demonstrated that two major factors might provide some explanation for the differences in utilization observed. These factors were distance from the service centres and level of income. They also examined "rurality" and failed to find any relationship between it and service take-up that could not be explained by distance from the service centre. Examination of physician distribution demonstrated that there was a largely urban bias in the distribution of family physicians, general practitioners, and obstetricians. Five rural census divisions had no physicians, either specialists or family physicians. In addition, the authors examined the patterns of referral by specialty, and were able to demonstrate that the Maritime provinces had large areas where no general or family practitioners referred for prenatal services and, even in areas with clinics, less than 20 percent of patients

were being referred. Analysis is being completed for the remaining Canadian provinces and for the country as a whole; it will be interesting to see whether similar patterns are repeated elsewhere.

Several factors might determine whether a particular woman is referred for prenatal testing. These include her knowledge of and desire for testing; her physician's awareness of the guidelines for testing and willingness to refer; personal biases concerning prenatal testing and its possible outcomes; and the perception of the possible risks involved. The survey of physicians by Chodirker and Evans (1993) in Manitoba, in connection with the maternal serum AFP screening program, shows that knowledge of the program, perceptions of its effect on the family, and personal biases affect individual physicians' willingness to involve their patients. In this study, several positive associations were identified, including a higher proportion of female physicians among those who referred all eligible women for prenatal counselling than among the group that did not. Urban Winnipeg physicians showed a greater knowledge both of prenatal testing and of maternal serum AFP screening than non-Winnipeg physicians. These observations tend to confirm that women living outside urban centres are likely to encounter difficulties in gaining access to prenatal services.

Recommendations:

Canadian medical schools should be encouraged to include among continuing medical education offerings courses on all aspects of the delivery and application of prenatal testing, including ethical implications.

Women's groups and others concerned about the health and well-being of women should be encouraged to develop objective and balanced informational modules about prenatal testing, including maternal serum AFP screening, which can be made available to women through their physicians' offices, public health units, antenatal classes, and other means appropriate to each particular region of Canada.

The provincial colleges of physicians and surgeons, and medical associations should emphasize to their members that failure to discuss with their patients the option for referral for a medically indicated prenatal diagnostic service is unethical and constitutes bad medical practice.

(cont'd)

Evidence from the Atlantic provinces suggests that lack of availability of obstetricians or family physicians in particular health districts may result in lower than expected utilization of prenatal services in these regions. If this finding can be shown to apply to the rest of Canada, then centres, in conjunction with their provincial health services, should try to ensure that at least someone in each public health or other appropriate health unit is knowledgeable concerning prenatal diagnosis and the options open to women at risk, so that women wishing such testing can obtain counselling near home and be referred to an appropriate centre for testing.

Genetic Counselling

Genetic Counselling Services

The delivery of genetic counselling services across the country varied with regard to both who counselled the prenatal patient and the format of the counselling session. In all genetics centres, medical geneticists counselled in complex cases and provided backup support and advice in other situations. The prenatal team was usually composed of one or more medical geneticists and prenatal diagnosis coordinators or genetic counsellors.

The role of the coordinator or counsellor showed tremendous variation across the country. In some centres they acted primarily to schedule and facilitate patient visits but, in most centres, they were also involved in prenatal counselling. The type of counselling performed varied between centres, from routine counselling when referral was for AMA to counselling for recurrent losses, consanguinity, previous aneuploid fetus or child, and molecular/biochemical diagnosis. Both the London and the Saskatoon centres used a team approach to prenatal counselling: the genetic counsellors took a pregnancy and family history before the woman was seen by the geneticist and spent time with her afterward. In almost every situation, the associate was the primary contact for the patient.

Maternal serum AFP counselling across the country exemplified some of the variations in prenatal counselling: in most centres where the referral rate was high and maternal serum AFP was in use (whether or not it was instituted as a formal screening program), most counselling was performed by a genetic counsellor. Where the test was sporadically or rarely done, the responsibility rested primarily either with the geneticist or with community physicians.

Most women undergoing prenatal testing did so for uncomplicated advanced maternal age. In many centres, counselling was done by the

community physicians and obstetricians; this was true in Halifax, Hamilton, Regina, Edmonton, Calgary, Vancouver, Victoria, much of Quebec, and some outreach centres. McGill triaged its prenatal referrals, dividing them into subgroups of "routine advanced maternal age patients" and "others." "Routine advanced maternal age patients" had the option of coming in for counselling or receiving a prenatal informational package in lieu of counselling. If the woman chose the latter, she completed and returned a family history form which was reviewed by the centre for anything unusual.

Another method used to facilitate counselling large numbers of women was the use of pregnancy questionnaires at the counselling session (Toronto General Hospital, Wellesley, and Calgary). Group counselling has been used in Calgary since 1991. The centres using pregnancy questionnaires did not take a formal pedigree unless a positive family history was elicited. In Halifax, Hamilton, and Vancouver, "routine advanced maternal age" referrals went directly to the obstetric unit where amniocentesis and chorionic villus sampling tests were performed.

The proportion of prenatal patients not seen by genetics personnel varied across the country; only one-third of the women undergoing invasive prenatal testing were seen by such personnel in St. John's and Vancouver. Quebec, Calgary, and Edmonton also had relatively high proportions of women seen only by obstetricians. In Winnipeg and Saskatoon, almost all women having testing were seen by a geneticist. Women who did not live close to a major centre, and where amniocentesis was available locally, were more likely to be counselled by an obstetrician. The exception was the provinces with outreach facilities, such as Ontario and Alberta. Almost all of the genetics centres attempted to facilitate access to prenatal diagnosis for women who had far to travel, either by phoning patients directly and discussing prenatal testing with them (patients were not formally counselled by phone) or by sending information about prenatal testing. In some centres, the geneticist would contact the woman's local physician to explain prenatal testing so that the information could be relayed to the patient. Most centres also tried to schedule the counselling and procedure for the same day, or over two adjacent days, so that only one trip was required.

All centres had unique characteristics that arose from the specific demands they had to meet, e.g., the Toronto Prenatal Diagnosis Program (TPDP) was a major referral centre for the Toronto metropolitan area for chorionic villus sampling and ultrasound scanning. Also, The Hospital for Sick Children was often sent referrals for "exotic" genetic disorders. Since the TPDP handled complex cases, this allowed centres like Wellesley to deal primarily with women at risk because of AMA or previous trisomy, serving chiefly the local Chinese and other ethnic communities.

The increasing reliance in some centres on written material in lieu of personal counselling, and the increasing number of Canadians whose first language is neither French nor English, suggests the need to have written

material and consent forms translated into the most frequently used languages.

Recommendation:

Centres with large immigrant populations in their catchment areas should ensure that written material and, in particular, consent forms are available in the most frequently used languages.

Approaches to Counselling: Patterns of Practice and Protocols

As previously mentioned, most genetics centres provided prenatal counselling either by medical geneticists or by genetic counsellors with appropriate backup. In centres such as Laval, University of Montreal (Ste. Justine), Toronto General Hospital, Credit Valley, Wellesley, London, Winnipeg, Calgary, and Edmonton, this was routinely done through established prenatal clinics. Saskatoon and Vancouver had no prenatal clinics per se but prenatal patients were admitted to combined general and prenatal clinics. In other centres patients were seen at a mutually agreed time.

The length of time for a routine AMA counselling session varied from 1.5 to 2 hours (St. John's, Saskatoon) to 20 to 30 minutes (Laval, Wellesley), with the average being about an hour. The number of patients seen each week also varied widely from about two per week (St. John's, Saskatoon, Victoria) to about 55 per week (University of Montreal, Toronto General Hospital, North York). The scheduling of counselling appointments was relatively uniform for centres offering amniocentesis, with most appointments booked for around 14 weeks' gestation. If chorionic villus sampling was also offered, a bimodal distribution was observed for the counselling sessions, with the earlier peak (around 8-10 weeks) corresponding to counselling for chorionic villus sampling. The risk most commonly discussed at the counselling session (for uncomplicated AMA) was that of having a live-born child with Down syndrome or any chromosome problem rather than the Down syndrome risk at the time of (Fetal loss rates are increased in chromosomally abnormal testing. This varied, however, depending on the specifics of the counselling session and the counsellor.

Partners and significant others were encouraged to attend counselling sessions and procedures; however, there were exceptions due to space or time constraints. For example, in Oshawa they could go only to the counselling session. Their attendance at an amniocentesis was not possible in North York or at McGill. All centres reported that additional counselling sessions would be available if requested by the woman or

couple. Very few of the centres sent letters directly to patients or copied doctors' letters for them. When this was done, it was usually because the case was complex and it seemed desirable to provide the family with a summary of what was discussed.

Across Canada the same general protocol was followed, with minor adaptations, for informing patients of abnormal results: the referring doctor was informed by telephone by a member of the genetics centre (usually the prenatal diagnosis coordinator or genetic counsellor). If the physician wished, this person would then inform the patient directly. Otherwise, the physician spoke with the patient, who was usually referred back to the genetics centre for further counselling. However, this was not necessarily the case. In Hamilton such patients were usually followed by the perinatologist. Patients of the TPDP were also not necessarily referred back to the genetics centre for follow-up counselling for standard trisomies of chromosomes 21, 13, and 18 because the physicians in the community were comfortable handling this. In Edmonton and Calgary, if patients were not initially counselled by the genetics unit, they were not necessarily referred for counselling.

Depending on the particular abnormality detected and the centre, the couple or woman was seen by a geneticist, a genetic counsellor, or both. The genetic counsellor was the main contact person, and kept in touch with the couple while they decided on a course of action. If the pregnancy was to be continued, the patient was referred back to the doctor with the understanding that the genetics centre would be contacted if further emotional support or information was needed (St. John's, Laval, London, Edmonton). Alternatively, the centre arranged for support services (McGill, Credit Valley, Vancouver). If the pregnancy was to be terminated, the centre either facilitated an abortion or referred the patient back to her physician. After a termination for genetic reasons, the woman or couple usually returned to the geneticist to review the pathological and chromosomal findings. A few centres (St. John's, London) forwarded the autopsy report to the patient's physician since recurrence risks were covered in the counselling session when the abnormality was first explained.

Information Available to Patients

In many of the centres, the use of aids such as video presentations was routine; exceptions were Laval, Ste. Justine, The Hospital for Sick Children, Regina, Saskatoon, and Victoria. These represent the smaller centres, one with a very specialized function (The Hospital for Sick Children), and most of the Quebec centres. Potentially, the lack of appropriate material in languages other than English may be a problem, both in Quebec and in large metropolitan areas where there are many recent immigrants.

All of the centres except St. John's, Ste. Justine, and Regina had prenatal pamphlets that dealt with the types of testing available and the

disorders these tests were generally used to detect. They did not cover in great detail the physical or mental handicaps of people with Down syndrome or neural tube defects, but these topics were usually raised in the counselling session, which presented an opportunity for them to be discussed in greater depth. Most of the centres gave out the information pamphlets at the time of counselling (except for patients who lived far away from the centre). Halifax, McGill, Wellesley, London, Winnipeg, and Vancouver centres routinely sent out pamphlets before the counselling appointment and, although the Ottawa service did not send out pamphlets before counselling, pamphlets were available in obstetricians' offices.

In all centres, when translation beyond the scope of centre staff was required, patients were requested to bring a friend or family member along to interpret. If this was problematic, translation services were available through the hospitals or from volunteer services. Language did not seem to be an insurmountable barrier to the delivery of genetic counselling services.

All centres maintained information about local, national, and international support groups for various genetic disorders. Such information was updated regularly. In 1991, the Canadian Association of Genetic Counsellors (CAGC) produced a compendium of genetic support groups available across Canada, and this has been supplied to each genetics centre in the country. In 1990, all centres maintained information about different disorders (e.g., journal articles and pamphlets produced by special interest groups) as a resource for couples. This information and any written information produced by the centre was updated as needed.

Invasive Prenatal Testing

Chorionic Villus Sampling

Availability

Only 12 centres in Canada offered chorionic villus sampling testing for prenatal diagnosis in 1990. In one, Calgary, it was available only for high-risk patients and not for AMA by itself (Table 3). Chorionic villus sampling was not available in Newfoundland, the Maritimes, Saskatchewan, or the territories.

At least 22 obstetricians involved with genetics centres performed chorionic villus sampling in 1990. Generally, it was performed by designated individuals. The highest number reported from a single centre was five, at Toronto General Hospital. Most centres offering chorionic villus sampling had one or two practitioners. The test was not offered in the community and, even in large urban areas such as Toronto, the service was concentrated in the larger teaching hospitals. Only one sample was reported as being sent into a centre for analysis from the community, the indication being risk for myotonic dystrophy.

Number of Women Having Chorionic Villus Sampling and Indications for Testina

Table 13 gives the number of women having chorionic villus sampling in 1990 and the number of procedures performed. In total, 2 097 women, or 9.4 percent of those referred to genetics centres, had the test. Relative to total referrals for prenatal diagnosis by region (Table 11), the highest proportion of referred women who went on to have the test was in British Columbia (27.1%); the lowest was in Quebec (2.1%). Rates were intermediate in Alberta (15.7%) and Manitoba (10.0%), and in Ontario the figures were higher for centres outside Toronto (12.8%) than within Metro Toronto (6.3%).

Table 14 gives the indications for having chorionic villus sampling performed. By far the highest number of tests were done for AMA (86.6%), although this number varied from 98.6 percent in Toronto to 66.1 percent in Quebec. The proportion was also relatively low in Alberta (76.9%) because Calgary did not perform the test for this indication. Compared to the total number of women referred because of AMA (Table 11), the older pregnant woman in British Columbia had the highest likelihood of having the procedure (1 in 3.7). The proportion of AMA patients tested tended to decrease from west to east: 1 in 4.6 in Alberta, 1 in 7.7 in Manitoba, 1 in 12.1 in Toronto, 1 in 7.4 in the rest of Ontario, and 1 in 53.6 in Quebec.

The next most common reason for chorionic villus sampling was having a previous child with a chromosome anomaly (7.5%). All 518 women referred for this reason elected to have invasive testing and 30.3 percent had chorionic villus sampling. It is likely that these women, conscious of their increased risk and having had prior experience of having a child with a genetic problem, more often presented early enough to consider chorionic villus sampling and sought the reassurance of an earlier diagnosis. However, a direct relationship seemed to exist between the general availability of the test and the proportion of these at-risk women who had it. Over 66 percent of those in British Columbia with this indication for testing had chorionic villus sampling, compared to 39 percent in Manitoba and Alberta, 35 percent in Ontario outside Toronto, 27 percent in Toronto, and 15 percent in Quebec.

Chromosomal anomalies in other relatives or in the parents accounted for 56 (2.7%) chorionic villus sampling tests. The proportion of such tests to total referrals was much higher for parental anomalies (30.8%) than for more distant relatives (4.0%), no doubt because of the generally low risks pertaining to the latter. The questionnaire data seemed to indicate that more prenatal tests were done for parental chromosome anomaly (138) than the number of women referred for this problem. It is unlikely the discrepancy is entirely due to repeat testing; it is probably because reasons for referral are more accurately recorded for patients subsequently having amniocentesis or chorionic villus sampling than for those who decline invasive testing. Regardless, the use of chorionic villus sampling among patients having invasive testing is higher for those with parental

Table 13. Number of Women Having Chorionic Villus Sampling Procedures in 1990

Region	Total no. of procedures*	No. of repeat procedures**	No. of women having the test
British Columbia	733		733
Alberta	284	3	281
Manitoba	101	4	. 97
Ontario (excluding Toronto)	461	10	451
Toronto	426	9	417
Quebec	118		118
Total	2 123	26	2 097

^{*} See Table 15.

chromosome anomalies (26.1%) than for those with other family members affected (10.4%). The regional trends observed earlier for chorionic villus sampling because of previously affected children are seen in these groups of patients.

Chorionic villus sampling was also widely used in two other groups of patients — those at risk for a child with an autosomal Mendelian disorder such as an inborn error of metabolism or chromosomal breakage syndrome and those at risk for a son with an undiagnosable sex-linked disorder. In 1990, the proportion of referred women having invasive prenatal testing for these conditions was 82 percent and 100 percent, respectively, with 24 percent of those with autosomal disorders and 50 percent of those requesting sex determination opting for the earlier test. Again, some discrepancies were found between reported indications for referrals for inborn errors of metabolism and the number of invasive tests performed, especially in Toronto and Quebec centres, which had relatively large numbers of such patients.

The Edmonton and Calgary centres were unusual in having 20 and five chorionic villus sampling tests, respectively, apparently performed after abnormal ultrasound findings. Vancouver also did four chorionic villus sampling tests for this reason as well as some for abnormal maternal serum AFP levels and for previous history of neural tube defects. It is unclear if this last indication was correctly reported, as chorionic villus sampling would not be the appropriate test in this circumstance. In the above centres chorionic villus sampling was occasionally performed in the second or even third trimester.

^{**} See Table 16.

Logistic Aspects of Chorionic Villus Sampling

The maximum number of chorionic villus sampling tests that could be performed per week varied from three in Ste. Justine to 16 in Vancouver. The main reason cited for restricting the number was lack of technological staff or because of other laboratory conditions. Occasionally, in smaller centres, chorionic villus sampling scheduling was difficult when the obstetricians involved were away. One centre, Vancouver, noted that limitations were occasionally placed because of lack of available appointments in the ultrasound facilities. No centre reported that counselling of patients was a problem; only the Ottawa service thought the demand was too low to create difficulties in offering chorionic villus sampling.

Different centres had different solutions to the problems of increased demand for chorionic villus sampling. In some centres, such as McGill, TPDP, London, and Vancouver, patients received priority on the basis of risk. In Calgary only high-risk cases were accepted. Other centres took patients on a first-come, first-served basis or gave priority to women from within their catchment area. Several centres kept waiting lists. In the two largest centres, women on the waiting list sometimes came into the unit on appropriate days and waited to see if an appointment became available due to a fetal death or other circumstance. Many centres reported giving preference to women closer to the upper gestational age cut-off for chorionic villus sampling or, if demand was excessive, they rescheduled patients to accommodate those at later gestations. Occasionally, centres would refer patients to other centres. However, it is likely that some women requesting chorionic villus sampling were informed that the procedure was not available for a variety of reasons. Amniocentesis was offered as an alternative in these situations.

In 1990, chorionic villus sampling was not performed in Canada before nine weeks' gestation. The upper limit varied, with most centres restricting the test to the first trimester. Some centres reported taking such samples later in pregnancy but did not document the reasons in detail. The median gestational age at chorionic villus sampling ranged from 10.0 weeks in British Columbia to 12.1 weeks in Manitoba.

From Table 15 it can be seen that the technique of choice for chorionic villus sampling varied across the country, with transabdominal chorionic villus sampling more common in Manitoba and Toronto and transcervical chorionic villus sampling in other parts of Ontario and in Alberta. Transcervical chorionic villus sampling was the only technique used in Quebec. Almost all centres reported that multiple pregnancies were considered a contraindication for chorionic villus sampling and that such patients would be recounselled and offered amniocentesis. In only three cases reported was chorionic villus sampling apparently done in a twin pregnancy, but the specific indications were not given.

		Total	1 000 (47.1)	746 (35.1)	377 (17.8)	2 123
	1	Quebec	118 (100.0)			118
	Ontario	Manitoba Toronto Excluding Toronto Quebec	432 (93.7)	19 (4.1)	10 (2.2)	461
		Toronto	79 (18.5)	303 (71.1)	44 (10.3)	426
sambling)			(10.6)	90 (86.5)		101
ionic Villus		Alberta	166 (58.5)	118 (41.6)		284
nscervical/total of chor		British Columbia Alberta	194 (26.5)	216 (29.5)	323 (44.0)	733
(% = ho. transabdominal or transcervical/total of chorionic Villus sampling)		Approach	No. of transcervical (%)	No. of transabdominal (%)	No. of unknown (%)	Total no. of chorionic villus sampling procedures

A total of 26 repeat chorionic villus sampling tests were performed and 134 women had an amniocentesis after a chorionic villus sampling test. Four women to whom it was not possible to give chorionic villus sampling results declined any further investigation. The most common reasons for repeat chorionic villus sampling (Table 16) were failure to obtain an adequate sample and failure to obtain a laboratory result. These were also common reasons for an amniocentesis after chorionic villus sampling (Table 17). However, many amniocenteses were done for investigation of an equivocal result after chorionic villus sampling or confirmation of an abnormal chorionic villus sampling test. Thus, 7.5 percent of women having chorionic villus sampling in 1990 required further testing. This is fewer than the 10.9 percent reported in the Canadian collaborative chorionic villus sampling trial (Canadian Collaborative CVS-Amniocentesis Clinical Trial Group 1989) and appears to reflect improved operator skill. There was no correlation between the number of repeat tests needed and relative frequency of transcervical versus transabdominal procedures. However, the frequency of repeat tests appeared highest in areas such as Manitoba (17.8%) and Quebec (13.6%), where few chorionic villus sampling tests were done, and lowest in centres performing many. Vancouver had only 2 percent of women requiring further testing but this may be an underestimation. The centre reported that repeat chorionic villus sampling was an option for patients but no such tests were reported. The follow-up of women after chorionic villus sampling varied between centres. In most

Table 16	Repeat Cho	rionic Villus	Sampling	by Region	(N)
Table 10.	nepeat ono	HOHIC VIIIUS	Samping,	by negion	(14)

		On	tario	
Alberta	Manitoba	Toronto	Excluding Toronto	Total
1	3	6	9	19
2	1			3
		2		2
		1	1	2
3	4	9	10	26
			4	4
	2	1 3	Alberta Manitoba Toronto 1 3 6 2 1 2 2 1	Alberta Manitoba Toronto Toronto 1 3 6 9 2 1 2

Table 17. Women Having Amniocentesis After a Chorionic Villus Sampling Procedure, by Region (N)

	Total	23	17	40	4	10	134
	Newfoundland*			-	-		2
	Quebec	12	2	v-	-		16
Ontario	Excluding Toronto	20	ю	4	ю		30
Ont	Toronto	14	ιO	10		ώ	37
	Alberta Manitoba Toronto	4	4	2	8	7	14
		2	ო	œ	7		20
	British Columbia	-		14			15
	Reason for amniocentesis	Failure to obtain an adequate sample	Failure to obtain a laboratory result	Equivocal laboratory result	Confirmation of abnormal result	Reason unknown	Total

Newfoundland and Saskatchewan did not have facilities to offer chorionic villus sampling in 1990. The woman from Newfoundland had chorionic villus sampling elsewhere and had a follow-up amniocentesis for an amnio assay to confirm the chorionic villus sampling result and also to confirm mosaicism detected on chorionic villus sampling. cases, an ultrasound scan was arranged for 16 to 18 weeks because chorionic villus sampling does not detect neural tube defects. In centres such as Toronto, Hamilton, Winnipeg, Edmonton, and Calgary where it was readily available, maternal serum AFP testing was also recommended.

Laboratory Aspects

A variety of techniques were used to culture chorionic villus tissue samples, especially for karyotypic analysis. Eight centres predominantly used long-term cultures, while direct preparations were used exclusively in one centre and in addition to long-term cultures by two others. In Calgary, samples from the first trimester were cultured long term, while those taken in late gestation to evaluate fetal anomalies were subjected to direct analysis. All laboratories required at least two slides to be evaluated independently before a result was reported, and 10 to 20 cells were considered the appropriate number for analysis. Most centres used G banding but Q banding was the preferred technique in London, Hamilton, and Winnipeg. All centres had similar protocols for dealing with suspected mosaicism. Usually as many cells as possible would be analyzed and ultrasound, amniocentesis, or cordocentesis offered depending on the finding (e.g., sex chromosome mosaicism, potential for a viable mosaic phenotype). Turnaround time from sampling to reporting of a result averaged 16 days but ranged from six days at Laval to 29 days at McGill.

Recommendations:

Given that chorionic villus sampling has been shown to be a relatively safe and reliable technique for first trimester prenatal diagnosis, centres should make this technique more widely available to women, especially those in high-risk groups, such as those who have previously had an affected child or who are at risk of having a child with a single gene disorder.

Chorionic villus sampling should be restricted to major centres where the number of tests performed will ensure that the obstetricians performing the test have sufficient experience. Centres offering this procedure should limit the number of operators to ensure that each has sufficient experience with the testing procedure.

Where chorionic villus sampling is medically indicated and not available in the local centre, provincial health care plans should meet both the cost of travelling to the nearest centre where the test is available and the out-of-province testing costs.

Amniocentesis

Availability

Twenty-two centres in Canada offered amniocentesis for prenatal diagnosis in 1990. Two of these, Regina and Victoria, acted as independent laboratories but used their main provincial centre to refer complex cases. Amniocenteses were also available in some outreach centres in several provinces and one territory (Table 2) and the fluids were shipped to the appropriate laboratory for analysis. Some tests were performed by obstetricians in community hospitals and again the fluids were shipped to the laboratories. Access to amniocentesis was most problematic for women in Prince Edward Island and the Yukon.

At least 138 obstetricians involved with genetics centres performed this test in 1990. In some centres, including Halifax, McGill, the Ontario centres, Winnipeg, and Vancouver, amniocentesis was carried out by designated individuals. In St. John's, Ste. Justine, Laval, Regina, Saskatoon, Edmonton, Calgary, and Victoria, samples were received from amniocenteses performed by several obstetricians in teaching or community hospitals. In these situations, patients were more likely to be tested nearer to their homes than, for example, in Manitoba where they usually had to travel to Winnipeg.

Number of Women Having Amniocentesis and Indications for Testing

Table 18 gives the number of women having amniocentesis in 1990 and the number of procedures performed. In total, 15 731 tests were done and potentially as many as 15 454 women had amniocentesis for prenatal diagnosis. Thus, approximately 70 percent of all pregnant women referred to genetics centres had an amniocentesis. Relative to total referrals for prenatal diagnosis by region (Table 11), the highest proportion of referred women who subsequently had amniocentesis was seen in Saskatchewan (89.5%): the lowest was in Alberta (56.2%). However, the numbers of referred patients are probably underestimated in Alberta, as the number of referrals noted for late maternal age are fewer than the number of amniocenteses and chorionic villus sampling actually performed for this reason. The figure of 56 percent probably reflects more accurately the proportion of activity of the genetics centres related to amniocentesis, given the significant involvement of community obstetricians in this testing. Rates were intermediate in British Columbia (71.9%), Toronto (70.0%), Newfoundland (69.7%), the Maritimes (68.6%), and Manitoba (66.2%). Quebec had a higher than average rate (79.7%), while Ontario outside Toronto had a relatively low rate (58.1%).

Obviously, in all centres the most common reason for amniocentesis was AMA (82.2%) (Table 19), although the percentage varied from 88.8 percent in British Columbia to 67.0 percent in Newfoundland. The proportion was also relatively low in Manitoba and Saskatchewan. Compared to the total number of women referred because of AMA (Table 11), such women in Newfoundland and Saskatchewan had the

highest chance of having an amniocentesis. Nearly all older women referred to prenatal diagnostic services in these provinces had amniocentesis. As the proportion of referred cases was low relative to the number of older pregnant women in the population in these two areas, it would appear they were not referred for counselling unless they intended to have testing or, more often, they declined to be referred. On the other hand, in Ontario outside Toronto only 60 percent of women referred for AMA subsequently had amniocentesis. All other provinces had intermediate rates. Unlike chorionic villus sampling, there were no obvious geographical trends.

It would appear that about 87 percent of older women referred had either amniocentesis or chorionic villus sampling. The numbers approached 100 percent in Newfoundland, Saskatchewan, Alberta, and British Columbia and varied from 83 to 89 percent in Manitoba, Toronto, Quebec, and the Maritimes. Only in Ontario outside Metro Toronto was the rate relatively low at 73 percent. To what extent these differences reflect the different referral patterns and record keeping in the centres and to what extent they are attributable to inherent differences in the women offered

testing by their primary physicians is difficult to assess.

The next most common reason for amniocentesis (3.9%) was an abnormal maternal serum AFP level. However, this indication was common only in Manitoba (17.2% of amniocenteses) and Toronto (8.5%) where over 50 percent of pregnant patients are screened by maternal serum AFP. Having had a previous child with a neural tube defect was a relatively common reason for amniocentesis in several centres, especially in Saskatchewan (5.9% of amniocenteses), the Maritimes (4.4%), and Newfoundland (3.2%) where the overall number of prenatal tests were low. However, in Manitoba, the proportion of amniocenteses for this reason was very low, reflecting the greater reliance in that province on maternal serum AFP screening and fetal assessment by ultrasound technology. Only 18 percent of women so referred in Manitoba had amniocentesis compared to nearly 100 percent in British Columbia and Alberta. All centres routinely did karyotypic analysis and biochemical testing in samples referred for abnormally high maternal serum AFP or a family history of neural tube defects.

Unlike chorionic villus sampling, an abnormal finding on ultrasound contributed to a relatively large number (3.6%) of the amniocenteses performed in 1990; this figure was especially high in areas such as Newfoundland (10.6%) and Saskatchewan (9.6%) where few tests were done.

Having had a previous child with a chromosome anomaly led to 2.3 percent of amniocenteses. As mentioned earlier, all women referred for this reason elected to have invasive testing and 69.7 percent had amniocentesis. The proportions having amniocentesis versus chorionic villus sampling

Table 18. Number of Women Having Amniocentesis in 1990

)			
Region	Total no. of amniocenteses*	No. of sets of twins with No. of repeat both sacs tapped amniocenteses**	No. of repeat amniocenteses**	No. of women having amniocentesis***
British Columbia	1 942	unknown	unknown	1 942
Alberta	1 034	. 10	15	1 009
Saskatchewan	272	4	ω	260
Manitoba	657	±∞	φ	641
Ontario (excluding Toronto)	2 107	21	47	2 039
Toronto	4 732	45	46	4 641
Quebec	4 440	=	39	4 390
Maritimes	453	2	4	447
Newfoundland	94	-	ω	82
Total	15 731			15 454

See Table 20. See Table 21.

This may be an overestimate as some centres returned incomplete information on the number of repeat amniocenteses and twin taps.

There was also one triplet pregnancy sampled with one amniocentesis per fetus.

Table 19. Indications for Amniocentesis Performed, by Region (% = no. referred for a particular indication/total of known indications)

Indication	British Columbia	Alberta	Saskatchewan	Manitoba
Advanced maternal age (%)	1 724 (88.8)	858 (83.0)	199 (73.2)	465 (70.8)
Previous chromosome abnormality (%)	26 (1.3)	24 (2.3)	11 (4.0)	7 (1.1)
Parental chromosome abnormality (%)	10 (0.5)	5 (0.5)	1 (0.4)	1 (0.2)
Relative with chromosome abnormality (%)	24 (1.2)	5 (0.5)	1 (0.4)	1 (0.2)
Abnormal maternal serum AFP (%)	22 (1.1)	10 (1.0)	6 (2.2)	113 (17.2)
Previous neural tube defect (%)	47 (2.4)	14 (1.4)	16 (5.9)	7 (1.1)
Inborn error of metabolism (%)	1 (0.1)	3 (0.3)	3 (1.1)	2 (0.3)
Other single gene disorder (%)	(0.2)	5 (0.5)		2 (0.3)
Disorder with chromosome marker/abnormality (%)	1 (0.1)			
Maternal/paternal irradiation (%)	2 (0.1)	3 (0.3)		
Abnormal ultrasound (%)	52 (2.7)	80 (7.7)	26 (9.6)	43 (6.5)
Teratogen exposure (%)	8 (0.4)	2 (0.2)	2 (0.7)	2 (0.3)
Sex for medical reasons (%)	3 (0.2)	3 (0.3)		
Ambiguous CVS result/failed CVS (%)	15 (0.8)	20 (1.9)	N/A	14 (2.1)

Table 19. (cont'd) (% = no. referred for a particular indication/total of known indications)

Indication	British Columbia	Alberta	Saskatchewan	Manitoba
Other indications*	4 (0.2)	2 (0.2)	7 (2.6)	
Unknown indications				
Multiply referred*	261	22	6	10
Total number known indications	1 942	1 034	272	657

^{*} Other indications for referral may not have been ones for which amniocentesis was an appropriate test.

directly related to the availability of the earlier test. Chromosomal anomalies in other relatives or in the parents accounted for 1.8 percent of amniocenteses. The proportion of amniocenteses to total referrals was much higher for parental anomalies (87.1%) than for more distant relatives (34.0%), again no doubt due to lower risks for the latter group. It should again be noted that more prenatal tests were done for parental chromosome anomaly (138) than there were women recorded as referred for this problem, probably due to record-keeping inaccuracies. However, it is apparent that use of amniocentesis among patients having invasive testing was lower for those with parental chromosome anomalies (73.9%) than for those with other family members affected (82.7%). Centres may have been more willing to consider the low risks in this situation a valid indication for amniocentesis but would perhaps have been less likely to offer chorionic villus sampling.

In two other groups of patients — those at risk for a child with Mendelian disorders and those at risk for a son with an undiagnosable sex-linked disorder — amniocentesis was widely used, especially where chorionic villus sampling was not available. These risk factors contributed a relatively small proportion (1.5%) of the total amniocenteses performed but were more common in Newfoundland (3.2%) where the number of prenatal tests was small. Quebec also had a relatively high proportion of amniocenteses for inborn errors of metabolism, perhaps reflecting the interests of the genetics groups there and the relatively high frequency of certain diagnosable disorders (e.g., tyrosinaemia) within the Quebec population.

•	Ontario				
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
49 (1.0)	56 (2.7)	38 (0.9)	1 (0.2)	4 (4.3)	161 (1.0)
		51			51
150	69	179	24	7	728
4 732	2 107	4 389	453	94	15 680

Rarer indications for amniocentesis were parental irradiation histories and teratogen exposure. The test was done only for 61.9 percent and 22.3 percent, respectively, of patients referred with these risk factors.

The final indication for amniocentesis was an ambiguous chorionic villus sampling result or failure to obtain a result after such a test. Again, this was closely related to the number of chorionic villus sampling tests performed. In British Columbia only 2 percent of 733 such tests were followed by amniocentesis, compared to Manitoba with 12.9 percent of 109 tests and Quebec with 13.6 percent of 118 tests being followed in this way. The centres with intermediate numbers of chorionic villus sampling procedures (Alberta, 284; Toronto, 426; the rest of Ontario, 461) had follow-up amniocentesis rates of 6.0 percent to 8.6 percent.

Logistic Aspects of Amniocentesis

Some centres reported that they could handle a maximum number of amniocenteses per week. This number ranged from 10 in Wellesley to 38 in Vancouver. However, 12 centres in different parts of the country stated that they had no specific limits. The centres that did restrict numbers stated that they had difficulty in scheduling procedures due to lack of obstetrical personnel or overcrowding of ultrasound facilities. Occasionally, centres noted that a shortage of technological staff or other laboratory conditions contributed to problems with providing this service, although this was much less commonly reported than with chorionic villus sampling. In general, centres like Toronto General Hospital, Winnipeg, Calgary, and Vancouver occasionally had problems with overload and shipped samples to other centres in Canada or the United States for analysis. Other centres coped with these situations by rescheduling patients at earlier gestations or occasionally referring the patients to other centres. There is little

evidence that women who are referred to the genetics centres and who request amniocentesis cannot have the procedure if it is medically indicated.

Amniocentesis may be less readily available, even in women with appropriate indications, when patients are referred at late gestational age, especially beyond 24 weeks gestation. All centres reported a willingness to counsel such patients and hopefully reassure them, and most would offer amniocentesis. However, all centres that would do amniocentesis in this situation noted that such women were counselled that termination of pregnancy, should an abnormality be found, was unlikely to be available.

Table 20 shows the distribution of amniocenteses performed by gestational age. Most (72.0%) were done between 16 and 19-6/7 weeks, while 24.0 percent were done at 13 to 15-6/7 weeks. It is unclear how many of these earlier amniocenteses were done before 14 weeks gestation, as many centres reported incomplete data for this variable. Both Quebec and Alberta reported experience with very early amniocenteses (< 13 weeks). A small proportion of amniocenteses were done after 20 weeks gestation and it is likely that at least a portion of these were due to abnormal ultrasound findings. The range of gestational ages through which centres reported they performed amniocentesis varied from nine weeks to 36 weeks. Only one centre, Calgary, would perform amniocentesis as early as nine weeks gestation; Edmonton, Saskatoon, and McGill would do the test between 11 and 13 weeks. The Calgary group has recently reported its experience with 400 amniocenteses performed before 15 weeks gestation (Iwanicki et al. 1992). Due to a high culture failure rate in tests before 11 weeks gestation, this is now their cut-off for offering the earlier Jørgensen et al. (1992) also found poor success in culturing amniocytes taken before 11 weeks. Across the country in 1990, the median gestational age at which amniocenteses were usually done ranged from 14 weeks in Calgary to 17.9 weeks in Regina. In 17 of the remaining 19 centres reporting these data, the median gestational age was in the sixteenth week.

Unlike chorionic villus sampling, amniocentesis was used widely in twin pregnancies, and over 100 amniocenteses were performed in which both sacs were sampled. In Manitoba one triplet pregnancy was also tested. The standard protocol in centres across the country was to recounsel women when twins were detected and to offer amniocentesis; however, this might entail the women travelling to a different hospital as not all obstetricians doing amniocentesis would do so for multiple pregnancies. Victoria noted that only one sac would normally be tested and Ste. Justine in 1990 also only tapped one sac unless fetal abnormalities had been detected. As of 1991, Ste. Justine taps both sacs. Data were not available from Vancouver.

A total of 173 repeat amniocenteses were performed, although this is a significant underestimation as data were not available from some centres. The most common reasons for a repeat amniocentesis (Table 21) were

failure to obtain an adequate sample and failure to obtain a laboratory result. Amniocentesis was rarely repeated to re-evaluate a previous amniocentesis result. In total, 1.1 percent of women having amniocentesis in 1990 required further testing by amniocentesis and at least two others had cordocentesis. This is much less than the 7.5 percent seen in patients referred for chorionic villus sampling. The frequency of repeat tests appeared highest in areas like Saskatchewan (3.0%) and Newfoundland (9.3%), where few amniocenteses were done, and lowest in centres performing many tests. The high frequency of culture failures in St. John's apparently occurred during one short period of time; these laboratory difficulties have not recurred.

One major difference between amniocentesis and chorionic villus sampling was that in many centres amniocentesis samples were sent to the laboratory from regional hospitals. The exact number of internal versus external samples was difficult to assess and, in some large centres, samples were sent to other laboratories when their demand was excessive. It is important to note that centres such as Halifax, Ste. Justine, McGill, Laval, and Regina that routinely received samples from the community rarely had difficulties with samples arriving without prior notification. In other centres when this occurred, the laboratory would usually set an amniotic fluid sample up if it arrived unexpectedly and then telephone the physician or hospital for more information. However, physicians were usually discouraged from sending samples without prior consultation.

Recommendations:

Given the provincial responsibility for the delivery of health care services, we recommend that interprovincial barriers be removed to allow each woman to receive prenatal testing in the most appropriate centre dealing with her particular problem. Accessibility would therefore not be denied if a local centre were unable to perform the testing.

Because the period between testing and the provision of results is a time of heightened anxiety for the woman, all centres need to be concerned about the length of time taken to provide results. It is our view that results from a second trimester amniocentesis for advanced maternal age should take no longer than three weeks, and results from a chorionic villus sampling for the same indication, no longer than two weeks.

Table 20. Number of Amniocenteses Performed at Various Gestational Ages

Gestation at which				
amniocentesis was performed	British Columbia	Alberta	Saskatchewan	Manitoba
Before 13 weeks		8	2	
13 to < 16 weeks	576	208	33	211
16 to < 20 weeks	1 048	266	91	278
20 to < 24 weeks	25	31	4	5
≥ 24 weeks	10	45	8	24
Unknown	283	476	134	139
Total no. of amniocenteses	1 942	1 034	272	657

^{*} McGill over-reported by 31.

Table 21. Repeat Amniocenteses, by Region (N)

	Alberta	Saskatchewan	Manitoba
Failure to obtain an adequate sample	1		
Culture failure/poor growth		7	5
Confirmation of previous amniocentesis result	1		1
Other		1	
Reason not given	13		
No. of repeat amniocenteses performed	15	8	6
Rate/100 amniocenteses	1.47	3.03	0.92

Note: No data were supplied by British Columbia.

Ontario					
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
1		42			53
552	606	996	169	46	3 397
3 618	1 326	3 258	269	28	10 182
63	67	138	6	7	346
12	45	37	6	8	195
486	63		3	5	1 589
4 732	2 107	4 471*	453	94	15 762

Ontario					
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
26	5	3			35
14	5	28	4	7	70
2	1	1		1	7
		1			2
4	36	6			59
46	47	39	4	8	173
0.98	2.23	0.89	0.89	9.3	1.27

Laboratory Aspects

Two main techniques were used to culture amniocytes for karyotypic analysis. Fourteen centres used *in situ* techniques while four reported using flask cultures. Calgary used flasks for early amniocenteses and *in situ* techniques for later gestations. In London, flask cultures were routinely used but *in situ* cultures were employed if the result was needed more quickly, such as in cases of fetal anomaly or where the gestation was advanced. The number of cells to be analyzed also varied. For *in situ* cultures, almost all laboratories required cells to be analyzed from more than one plate, although two did not specifically state this. The number of colonies usually assessed varied from two to 10, and 10 to 30 cells were normally analyzed. All but one centre using flask techniques required at least two flasks with eight to 20 cells per flask analyzed. Most centres used G banding, but Q banding was the preferred technique in Hamilton, London, and Winnipeg.

Centres varied in their protocols for dealing with suspected mosaicism. Usually, as many cells as possible would be analyzed, but if level III mosaicism was suspected it was either reported as such (eight centres), a cordocentesis was offered (eight centres), or a repeat amniocentesis was suggested (four centres). Usually, further counselling and fetal assessment

were offered to the family.

With respect to biochemical assessments, amniotic fluid AFP levels were routinely evaluated in patients undergoing amniocentesis. However, the protocol for further evaluation with acetylcholinesterase varied between centres. Cut-offs for high amniotic fluid AFP leading to testing ranged from 2.0 multiples of the median (MOM) or +2.0 standard deviation (SD) to +3.0 SD. Other centres reported that samples from all amniocenteses with elevated maternal serum AFP were also sent.

Turnaround time averaged 21 days across the country, ranging from 12 days to 38 days. Generally, centres analyzing a small number of tests, such as St. John's, Regina, and Saskatoon, had a result available in a shorter time.

Recommendation:

Laboratory techniques used for prenatal diagnosis and the protocols used to evaluate unusual results, including mosaicism, are variable. The Cytogenetics Committee of the CCMG is in the process of establishing guidelines in this area as to the best way to approach this evaluation. All centres should be encouraged to apprise themselves of this information and follow recommended protocols when they have been finalized.

Cordocentesis

A total of 186 cordocenteses were reported as having been done in a total of 10 centres. This test was used in all provinces except Prince Edward Island, New Brunswick, and Newfoundland. However, the proportion of referred women having cordocentesis varied considerably from 1 in 24 in Halifax to 1 in 172 in Quebec. Only in Halifax, Manitoba, and Saskatchewan did the rate exceed 1 in 100. Not all centres, including Halifax, documented specifically the indications for cordocentesis. The centres identified 28 physicians who were doing this test, and the number available per centre varied from four in Vancouver to one in Toronto General Hospital.

Among known indications, the most common reason (82%) was fetal abnormality or other complication noted on ultrasound. This included intrauterine growth retardation, fetal malformation, and hydrops. However, in Manitoba 46 percent of cordocenteses were done for low maternal serum AFP indicating possible chromosome anomaly or other problems. These women were determined through the screening program as being at increased risk and offered this test as their gestations were relatively far advanced. At least two tests were done to obtain fetal blood for specific studies (risk of fragile X and haemoglobinopathy, respectively) where the diagnosis presumably could not be made adequately from cultured cells. Only two tests in 1990 were reported as having been done to re-evaluate the results of an amniocentesis. Two other reasons given for cordocentesis deserve comment. This test was used in at least three cases where oligohydramnios was present and presumably offered as an alternative to amniocentesis. One other test was done to evaluate a fetal death. Investigation of fetal death was rarely reported as a reason for prenatal testing in this survey. However, it should be noted that cytogenetic evaluation is more likely to be successful in cases of fetal death if samples are collected by invasive techniques rather than attempting analysis on fetal cells after the fetus is delivered (Brady et al. 1991). This may make definitive diagnosis possible and it has implications for subsequent genetic counselling.

Centres that provided information on the subject noted that they generally quoted risks of 0.5 to 2 percent of fetal loss after cordocentesis but that this could be higher in already compromised fetuses. We are aware of one fetal loss in this sample of 186 cases but outcome data are incomplete.

Eligible Women Not Having Invasive Prenatal Testing

Without more precise data it does not seem possible to determine with accuracy the factors that influence use of invasive tests (especially by older women) across the country. Probably, the availability of amniocentesis and chorionic villus sampling, distance to be travelled, and local physician and patient attitudes all play a role. As far as we have been able to determine, approximately 9.7 percent of patients referred for amniocentesis or

chorionic villus sampling declined the procedure (Table 22). This varied from 20.4 percent in Ontario centres outside Toronto to 3.7 percent in Halifax. Newfoundland (17.3%) and Toronto (10.1%) also had relatively high refusal rates; rates for Saskatchewan (7.7%), Manitoba (6.1%), Alberta (4.6%), and Quebec (4.4%) were lower. No data were available from British Columbia. As can be seen from the absolute number of women placed in the refusal category, these data must be interpreted with caution, as record-keeping differences between centres will have influenced their accuracy. However, there is some indication that, in areas where community obstetricians play a large role in the delivery of prenatal diagnostic services, the women who are referred to the genetics centres usually have decided in advance to have invasive testing. The high refusal rates in Ontario may indicate that, when women do not have as far to travel for counselling and testing, they may be more likely to decline after counselling.

In some cases, invasive prenatal testing is requested by the patient but is not done because a fetal death is observed on ultrasound examination immediately before testing or the patient miscarries before the test appointment. Again, these data seemed to have been collected with different degrees of accuracy by the centres and therefore may be potentially underestimated. The proportion of dead fetuses found at amniocentesis or chorionic villus sampling varied from none in Manitoba (probable under-reporting) to 3.3 percent in the Maritimes. The figures in most centres were low, especially in areas where prenatal testing is done in the community in many cases. Many such women likely do not come to the attention of the local genetics centre. A higher proportion of women were known to have miscarried before a test; this ranged from 6.3 percent in Ontario outside Toronto to apparently none in Newfoundland. Again, it is likely that only women who were directly counselled and had test appointments arranged by the genetics clinics are likely to be determined as having had a miscarriage before testing. It would be valuable to know what kind of follow-up investigations and counselling are offered to such women.

Non-Invasive Prenatal Testing

Ultrasound

In all but two centres, diagnostic ultrasonographic examinations requested by genetics centres were performed in fetal assessment units affiliated with the local teaching hospital or a hospital attached to the centre. In centres where such examinations were done in other units, a written report was sent to the genetics centre. Centres varied slightly in their choice of a gestational age when such examinations would be performed; this ranged from 15 to 20 weeks with a median of 18 weeks.

Indications for genetic ultrasound scans are given in Table 23. This table does not include post-chorionic villus sampling examinations or those

carried out before a chorionic villus sampling or genetic amniocentesis. The most common reason for referral for ultrasound scanning was an increased risk of structural abnormality in a fetus. It is not possible to relate the numbers directly to total referrals but centres apparently varied in the frequency of use of ultrasound for this purpose. For example, Hamilton had 394 such referrals compared to only 63 in Ottawa, despite similar population bases in the two areas.

The next most common reason for ultrasound scanning was an abnormal maternal serum AFP level; however, this was used only in the centres that had relatively routine maternal serum screening. In Manitoba, 448 women had a fetal assessment in 1990 for abnormal maternal serum AFP results. However, these scans were arranged by the Manitoba Maternal Serum AFP Screening Program coordinator rather than coming directly from the Section of Clinical Genetics.

At least 137 ultrasound examinations were done for women who had declined invasive procedures, presumably for reassurance and/or to attempt to identify any physical signs suggesting Down syndrome. At least 12 of the 98 scans done for other reasons were also documented as being done for maternal anxiety or having had a previous child with Down syndrome. However, several centres, including North York, Credit Valley, and Winnipeg, believed that this was an inappropriate referral and would offer maternal serum AFP screening instead. Although there have been reports in the literature suggesting that certain signs, such as nuchal thickening and femur length, may be useful in identifying fetuses at risk of Down syndrome (Benacerraf et al. 1987; Bouchard and Bissonnette 1989; Brumfield et al. 1989; Toi et al. 1987), these studies remain controversial and the sensitivity and specificity of such screening are not well established. If women are likely to be given a false sense of security by apparently normal ultrasound screening despite counselling to the contrary, it may not be appropriate to recommend ultrasound in such cases. The only other common indications for ultrasound scanning were a previously abnormal ultrasound or exposure to teratogens.

Recommendation:

The CCMG and SOGC should evaluate the practice of using ultrasound screening to rule out chromosome abnormalities, in particular Down syndrome, and should prepare guidelines to ensure that such ultrasound screening is applied appropriately.

Table 22. Reasons for Not Having an Invasive Test

			_
	Alberta	Saskatchewan	Manitoba
No. of women declining invasive testing	64	22	50
Dead/disorganized fetus found at time of test	2	1	
No. of women miscarrying before testing	46	1	22
Total	112	24	72
No. of women having amniocentesis or chorionic villus sampling	1 270	260	724

^{*} Includes 2 660 women from British Columbia: no data on reasons for not having invasive tests were supplied by that centre.

Table 23. Number of Women Referred for Ultrasounds Through Genetics

	Alberta	Saskatchewan	Manitoba
Requested by Genetics because of a maternal serum AFP result		5	2*
Fetus at increased risk of structural malformation	62	75	58
Amniocentesis/chorionic villus sampling declined by patient	8	10	15
Ultrasound for other indications	11		27
Total	81	.90	102

Most ultrasounds requested for abnormal maternal serum AFP values are arranged by the maternal serum AFP coordinator and not by Genetics. In 1990, 448 women had ultrasounds for abnormal maternal serum AFP values.

Note: No information was returned by British Columbia, Credit Valley, Wellesley, and Regina.

Ontario					
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
594	695	222	18	18	1 683
44	4	33	16	. 1	94
173	213	276	5		731
811	912	531	39	19	2 508
5 021	2 460	4 492	447	84	17 418*

Ontario					
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
154	83				244
460	511	. 540	16	43	1 765
23	46	27	8		137
44		15	1		98
681	640	582	25	43	2 244

Maternal Serum AFP Screening

Availability

Only in Manitoba was maternal serum AFP available as part of a provincial screening program. However, both the Oshawa centre and Toronto General Hospital indicated that screening was offered routinely in their local population. As far as these centres were concerned, all their prenatal genetics patients had maternal serum AFP screening, but other pregnant women may or may not have been screened depending on their private physician. Some physicians screened all patients, while others restricted the test to high-risk patients. The remaining centres reported that maternal serum AFP screening was not done as a population screen, although Credit Valley, Hamilton, London, and Vancouver reported that some physicians screen routinely. In Regina, screening was done by private physicians but the local genetics centre (primarily a cytogenetics service facility) was not involved in following up these women.

All centres had the capacity to do maternal serum AFP evaluations for appropriate patients. With respect to indications, many centres, including Wellesley, North York, and London, screened all prenatal genetics patients. In 1990, St. John's also started screening genetics patients to determine local normative maternal serum AFP values. Ste. Justine, McGill, Hamilton, Ottawa, Edmonton, and Vancouver requested samples on all post-chorionic villus sampling patients. All centres that did not screen routinely offered maternal serum AFP testing to those with a family history of neural tube defects and most also used maternal serum AFP to evaluate women with diabetes. Exposure to teratogens, especially Valproic acid, and an abnormal ultrasound examination also led to maternal serum AFP screening in some centres. Maternal anxiety and, to a lesser extent, family history of Down syndrome were common indications for referral in some centres.

Logistic Aspects of Maternal Serum AFP Screening

Most centres recommended screening be done around the sixteenth week of pregnancy. The lower limit of gestational ages acceptable for samples ranged from 14 to 16 weeks with a median of 15 weeks. The upper limit ranged from 16 to 24 weeks with a median of 18 weeks. Only the Manitoba program would routinely screen patients beyond 20 weeks gestation; the program's recommended time for screening was 16 to 18 weeks.

Seven centres used normative values derived from local populations; seven used standards provided by the manufacturer of the test assay kit. Two centres used a combination of local and kit data. Calgary was developing normal curves for early amniocentesis patients and Saskatoon was also developing local standards. Ottawa and Hamilton noted that they used standards developed by a provincial reference laboratory. Edmonton

sent all its samples to Winnipeg for testing and interpretation, and Victoria's samples were handled in Vancouver.

Cut-offs for further evaluation of elevated maternal serum AFP levels ranged from 2.0 MOM to 2.5 MOM. Eleven centres used 2.5 MOM and three used 2.0 MOM. Ottawa used 2.5 MOM, but has since shifted to 2.0 MOM. The Manitoba program used 2.3 MOM. There was more variability in the cut-offs used for low maternal serum AFP indicating increased risk for Down syndrome. Five centres used a MOM cut-off ranging from 0.5 to 0.7, with three using 0.5 MOM. Six centres used an age-equivalent risk of a 35-year-old. Credit Valley and Hamilton used a combination of offering amniocentesis to those with high age-adjusted risks and to younger women with values below 0.5 MOM. The Quebec centres and Ottawa did not usually report maternal serum AFP age-adjusted risks for Down syndrome. In Manitoba, in 1990, the age-adjusted risk cut-off was equivalent to that of a 37-year-old, but this has subsequently been changed to that of a 35-year-old.

Follow-up of patients with high maternal serum AFP levels usually included a combination of counselling, fetal assessment, and amnio-The precise protocols varied between centres. Six centres reported they counselled patients before ultrasound examinations were done, three noted they counselled after an ultrasound had been done, and six did not mention counselling specifically in their follow-up protocol. Only two centres reported they got repeat maternal serum AFP samples on such patients. Only one centre, Winnipeg, noted that amniocentesis was not routinely used to evaluate such patients but that they had a repeat fetal assessment two weeks later if no reason for elevated maternal serum AFP was detected. Oshawa also noted that a second ultrasound would be done at 20 weeks if a patient declined amniocentesis. Follow-up of patients with unexplained elevations of maternal serum AFP was not commonly done by the genetics centres, but Ottawa and Vancouver noted specifically that this would be arranged by private physicians. Calgary kept in touch with the patient until delivery and Ste. Justine would arrange to evaluate the baby after birth. In Winnipeg, women were followed up every four weeks with repeat fetal assessment, maternal serum AFP, and Kleihauer Edmonton followed up patients with repeat ultrasounds and maternal serum AFP, and Toronto General Hospital also used serial ultrasound examinations for such patients.

Patients with low maternal serum AFP at an increased risk of Down syndrome were usually counselled and offered amniocentesis if ultrasound examinations confirmed their dates. Calgary offered triple testing first and offered amniocentesis only to those remaining at increased risk. In most centres, if the results of the amniocentesis were normal, these patients were not followed. However, in London, a repeat ultrasound would be arranged if the patient declined amniocentesis. As mentioned, the Quebec centres and Ottawa did not report Down syndrome risks, but the Ottawa

service would offer amniocentesis to women with low maternal serum AFP if they were 33 or 34 years of age. If they were younger a dating scan would be recommended. Regina had no protocols for following up patients with high or low maternal serum AFP values; this was managed by the physicians ordering the test.

All centres were aware of the need to arrange counselling quickly for women with abnormal maternal serum AFP levels. Such patients were scheduled when possible into regular prenatal clinics but would be counselled at other convenient times as needed. In Winnipeg, the Manitoba maternal serum AFP program coordinator saw almost all of these patients and they had the opportunity for fetal assessment and amniocentesis, if required, immediately afterward. In other centres, patients would be counselled by either geneticists or genetic counsellors, and amniocentesis, if requested, was done within a week at the most.

Centres varied in whether or not they routinely did maternal serum AFP testing in patients already undergoing amniocentesis. Nine did and five did not. Ste. Justine and McGill drew samples, but they were not analyzed unless the amniotic fluid AFP was elevated. Calgary drew samples only from its early amniocentesis patients. Only centres that had preamniocentesis maternal serum AFP results could come across the situation of a woman who had normal amniotic fluid AFP (largely ruling out an open neural tube defect) but an elevated maternal serum AFP. Toronto General Hospital and the Manitoba program, both of which routinely followed patients with unexplained maternal serum AFP levels, followed these patients in the same fashion. London also followed these patients with repeat ultrasound. Other centres did not follow such women specifically, although in Halifax and Ottawa the private physician would be alerted to watch for intrauterine growth retardation and other complications.

Number of Screened Patients

Table 24 provides information on the number of women screened with maternal serum AFP in 1990, the number ascertained with an abnormal value, the number of counselling sessions, and the number of amniocenteses performed. With the exception of the Manitoba program, the figures reflect only those patients known to the genetics centres; however, there may be a significant number of women being screened in the community with little, if any, genetics centre involvement, even when an abnormal test result occurs. Only Manitoba, Toronto General Hospital, North York, and Halifax stated that written material on maternal serum AFP screening was readily available in doctors' offices. Some other centres included information on AFP in material provided to prenatal patients.

The largest number of screened patients occurred in the Toronto area, followed by Manitoba. Screening was uncommon in Quebec. The proportion of women with an abnormal value was 7.5 percent across the country, ranging from 2.0 percent in British Columbia to 29.4 percent in Newfoundland. High percentages of abnormal results were seen in centres

where screening was used only for high-risk cases, with the exception of Quebec. However, abnormal results in Quebec related only to elevations of maternal serum AFP. The reason for the low number of abnormal maternal serum AFP values in Vancouver is hard to determine, but the number of screened women was reported only as an estimate. Since all women reported as having abnormal AFP values were counselled, it is likely that many such women were followed up in the community and not reported to the genetics department. Counselling for abnormal AFP values was more common in Manitoba (81%) than in Toronto (33%). This is probably because in Oshawa, Credit Valley, and North York women are counselled only after an ultrasound has been done; thus, they may not be referred if a reason for an abnormal result is detected.

As far as we have been able to determine, 186 amniocenteses were performed for high maternal serum AFP. For the 41 in which maternal age was known, 13 women were already eligible for amniocentesis because they were 35 years of age or older. At least 10 had previously declined amniocentesis based on late maternal age. At least 469 amniocenteses were done because of low maternal serum AFP indicating increased risk of Down syndrome. Of the 310 of known age, 102 were over 34 years and at least 43 had previously declined amniocentesis. Toronto General Hospital noted that 65 percent of women offered amniocentesis because of low maternal serum AFP, indicating increased risk of Down syndrome, declined. In the Manitoba program the figure was 62 percent for women of AMA and 37 percent for those under 35 years of age. The ratio of amniocenteses for low versus high maternal serum AFP was significantly higher in Manitoba. Toronto General Hospital also noted that most (85%) of their patients with high maternal serum AFP declined amniocentesis after fetal assessment showed no apparent fetal abnormality.

Recommendation:

Given the different protocols used to offer maternal serum AFP screening across the country, the test should be offered only on a population basis, within the confines of a program that adheres to the guidelines established by the American Society of Human Genetics (American Society of Human Genetics 1987; Garver 1989) and affirmed by the CCMG (Davidson 1987). Where the resources to develop such programs and the associated counselling are not available, the test should be restricted to patients at high risk.

Triple Testing

Only one centre, Calgary, indicated that triple testing — using a combination of age and levels of maternal serum AFP, beta human

Table 24. Maternal Serum AFP Testing Within Centres Offering Prenatal Diagnosis, by Region

(% = abnormals/number of pregnant women screened)

	British Columbia	Alberta	Saskatchewan	Manitoba
Is there a provincial screening program?	no	no	no	yes
No. of pregnant women screened with maternal serum AFP	1 300	889		9 267*
No. of women determined with at least one abnormal maternal serum AFP value (%)	26 (2.0)	45 (5.1)		696 (7.5)
No. of counselling sessions for maternal serum AFP	26	10		561
No. of amniocenteses for high maternal serum AFP	5	4	6	13
No. of amniocenteses for low maternal serum AFP	15	6		100

^{*} The figures given for Manitoba are from the provincial Maternal Serum AFP Screening Program and include all women screened in the province and not just those screened through the genetics department. Samples from Alberta, which were sent to the Manitoba Maternal Serum AFP Screening Program for analysis, and women screened at the time of amniocentesis are not included.

chorionic gonadotropin, and estriol to assess Down syndrome risks — was being used in 1990. This was part of a pilot program and was offered to women attending the prenatal diagnosis clinics and to those with low maternal serum AFP levels. In 1991, Winnipeg began a study to collect normative data but has not yet begun to report individual patient risks.

Follow-Up and Outcomes of Pregnancies in Prenatal Diagnosis Patients

Mechanisms

With one exception, all centres reported that they had a formal review process or audit, the frequency of which varied from monthly to annually. The issues discussed at these meetings usually included the number and type of procedures performed, the indications for prenatal diagnosis, and

^{**} This does not include Hamilton.

Ontario Toronto Excluding Toronto		Ontario			
		Quebec	Maritimes	Newfoundland	Total
no	no	no	no	no	
22 109	3 384**	134	63	17	37 163
1 836 (8.3)	174** (5.1)	9 (6.7)	10 (15.9)	5 (29.4)	2 801 (7.5)
596	98			5	1 296
142	16		1		186
300	42		6		469

abnormal results. Not all centres had the opportunity to review pregnancy outcomes, especially those of women with normal test results.

The process for collecting information about pregnancy outcomes varied considerably among centres. However, only three centres reported that they did not routinely get information on outcomes and one other followed up only high-risk cases. The information was gained primarily through initial contact with the patient in nine centres and from the referring physician in six others. Two others used mostly hospital chart reviews. Credit Valley and Winnipeg had prenatal patients sign forms for release of medical information at the time of counselling, and London gave the patient a follow-up form at the counselling session to be returned at the appropriate time. The follow-up of patients screened by maternal serum AFP also varied. In several centres where screening was offered only to patients through the genetics programs, the women were followed up in the same way as other prenatal patients. In Ontario, where community-wide

Terminations of pregnancy for genetic reasons were usually followed up with a detailed evaluation of the fetus and often confirmation of the results by amniocentesis at the time of induction or culturing of fetal tissues. Follow-up of spontaneous abortions was less routine. Often, centres were not aware of such events unless informed by the patient or her physician or until outcome data were routinely collected. Information was then reviewed to evaluate the cause and timing of the fetal loss.

Although several provinces, including Nova Scotia, Ontario, Manitoba, Alberta, and British Columbia, had registries recording births of infants with congenital malformations, only Manitoba and British Columbia had a formal system to link data in the registry with the prenatal diagnosis and genetics programs.

Options Available to Women with Abnormal Results

Many women, faced with the knowledge that their fetus has a major handicapping or lethal condition, seek a termination of their pregnancy: however, others prefer to continue. The options available to women considering prenatal diagnosis were usually discussed at the time of counselling in genetics centres. Therapeutic abortion for women with abnormal results determined in the first trimester was available in all centres and was done as an outpatient procedure in all but one centre. In the second trimester, when most results became available, terminations were more restricted, especially in the Maritimes. In St. John's, such terminations were available only for lethal disorders such as an encephaly. However, only Vancouver reported that terminations of pregnancies for genetic reasons in the major hospitals were approved through abortion committees in 1990. Termination of pregnancy usually occurred within seven days of diagnosis (median four days) and was available until 20 to 24 weeks (median 22 weeks) for non-lethal anomalies. Pregnancies with lethal anomalies could usually be terminated at any time. Second trimester terminations were inpatient procedures and many centres tried to admit such women onto gynaecological rather than labour and delivery wards, and often into private rooms.

The number of obstetricians willing to perform second trimester terminations for genetic reasons varied from two in Oshawa to over 100 in Toronto, but they were not performed by other physicians. In rural areas, women would likely have to travel some distance to receive a termination

of pregnancy, incurring considerable inconvenience and, especially if they had to go out of province, personal expense.

Only one centre thought that prenatal testing was inappropriate if a woman was emphatic that she would not have a termination of pregnancy for any reason. Other centres considered the mandate of prenatal diagnosis was to provide information to the couple and that their decision of what to do with the information was a separate issue.

All centres noted that they offered counselling to women when an abnormal result was obtained. In some centres where most pretest counselling was done by community obstetricians, such offers were less often accepted and the local obstetricians were responsible for arranging pregnancy terminations when requested. Although most centres noted that the geneticists would be primarily involved in such counselling (often the person who had first seen the family), three said that they involved families who had had similar experiences in the past as additional resources for parents. If families elected to continue the pregnancy, the genetics centre would arrange to see the infant on consultation after delivery, would offer additional counselling, and would facilitate referrals to community services and support groups. If the woman decided to terminate the pregnancy, follow-up counselling would also be offered. Several centres involved social work, pastoral care, or psychological support services in the management of such patients, and all offered counselling to review all results and to re-evaluate recurrence risks if the family wanted. Often, genetic counsellors had a major involvement in the follow-up and support of such patients.

Outcome Information Available

Centres were requested to provide information on abnormal results obtained by invasive and non-invasive prenatal testing, and on the outcome of such pregnancies, abnormal outcomes of pregnancy in women with normal test results, and known false positive and false negative results.

Sixteen centres returned some outcome data for patients seen in 1990. For three centres, the only information available was for abnormal cytogenetic results; six other centres provided relatively complete data but only for patients with abnormal test results. Seven centres gave information on outcomes in women with both normal and abnormal results.

Recommendations:

All centres should declare as explicit policy that agreement to terminate a pregnancy is not a precondition or requirement for undergoing prenatal testing.

(cont'd)

All Canadian women, wherever their location, should have reasonable access to prenatal testing and be aware of the options open to them after learning the test results. Those opting for termination of pregnancy should be provided with the necessary referral to achieve that option and should not normally have to travel out of province to obtain the service.

Where they do not already exist, all centres providing prenatal testing should have, within their centre or by referral, facilities to provide women and their partners with both pre- and post-termination counselling, including grief counselling.

Abnormal Results

Cytogenetic

We have results on cytogenetic abnormalities from 16 centres. A total of 1 206 women had chorionic villus sampling and 10 481 had amniocentesis. The number and frequency of anomalies reported by these centres are shown in Table 25. This must be considered a minimal estimate as not all centres may have reported all balanced or variant anomalies. However, it would appear that a woman having an invasive test had a 1 in 29 chance of having some chromosomal change documented in the fetus.

The most common single anomaly reported was Down syndrome. The most common indication for testing in these cases was AMA, although 10 percent had been tested because of an abnormal ultrasound and 5 percent because of low maternal serum AFP. Of those pregnancies where outcomes were known, 87.6 percent elected to terminate the pregnancy, 4.5 percent had spontaneous losses, and 7.9 percent had live births (one infant died in the neonatal period). Trisomy 18 was seen in 49 cases, and a higher proportion (20.4%) of these women had been tested because of an abnormal ultrasound examination. Of the known outcomes, 73.8 percent ended in termination and 9.5 percent in live births. The other women had spontaneous abortions or stillbirths. Eight fetuses with trisomy 13 were identified, six in mothers of AMA and the other two because of abnormal ultrasounds. Seven pregnancies ended in termination and one in a live birth. The natural history of these conditions includes a higher than usual spontaneous loss rate during pregnancy and a greater likelihood of stillbirths.

Sex chromosome anomalies, especially 45,X in pure or mosaic form, were identified in 71 fetuses. In a third of the Turner syndrome cases, the diagnosis was made after an abnormal ultrasound. Two others had low

Table 25. Cytogenetic Anomalies Reported

	No. reported	Rate/1 000 women having invasive tests
Down syndrome	100	8.56
Trisomy 18	49	4.19
Trisomy 13	8	0.68
Sex chromosome anomalies 45,X and mosaics 47,XXX 47,XXY 47,XYY	51 7 10 3	4.36* 0.59* 0.86* 0.26*
Other numerical anomalies	36	3.08
Structural anomalies Robertsonian translocations Reciprocal translocations Inversions Other structural anomalies	13 38 15 12	1.11 3.25 1.28 1.02
Variant chromosomes	57	4.88
Total	399	34.12

^{*} Rates not corrected for fetal sex.

maternal serum AFP. In 70.5 percent of these cases with known outcomes, termination of pregnancy occurred. Not unexpectedly for this condition, where the majority do not survive to birth, 46 percent of the continuing pregnancies ended in spontaneous abortion or stillbirth. Four of the seven pregnancies that went to term were of mosaic 45,X/46,XX fetuses. There were four fetuses with karyotypes of 45,X/46,XY; two ended in termination and the outcome of the others is not known. Other sex chromosome anomalies seen were Klinefelter's syndrome, XYY, and triple X. The reason for referral was almost always AMA. In over 70 percent of the cases where an outcome is known, the pregnancy was carried to live birth.

Other numerical chromosome abnormalities included marker chromosomes and mosaicism for a cell line with a missing or extra chromosome. Four of the 14 mosaics involved chromosome 20. Termination of pregnancy was chosen in 50 percent of the cases with markers and 33 percent of those with mosaicism. Of the five cases with additional markers or an extra chromosome in a mosaic form, all ended in the births of apparently healthy newborns. The three cases with mosaicism for missing chromosomes ended in premature delivery at 27 weeks in one instance and, in the other two, with the births of children with severe heart

defects. Ten triploid fetuses were identified, three after morphological anomalies were detected on ultrasound.

Among structural chromosome rearrangements, Robertsonian translocations were less common than reciprocal. This may be an ascertainment bias, as prenatal testing may be less commonly recommended in cases of Robertsonian translocations not involving chromosomes 13 or 21. The data provided were not complete but it would appear that at least four fetuses had unbalanced rearrangements due to parental translocations and these were all terminated. At least nine of the translocations detected were *de novo* (i.e., not carried by either parent). In five of the seven cases with known outcomes, the pregnancies were continued. Although parental chromosomal defects were the reason for testing in some of the cases with translocations, a more common reason was AMA, and the translocations were unexpected findings.

Late maternal age was also the usual reason for referral in most of the women with other structural rearrangements. In all cases where the outcome is known, the pregnancies were continued when the anomaly was a balanced inversion or insertional translocation. However, in six cases with unbalanced karyotypes such as deletions or duplications, the pregnancy was terminated, and, in one other, the infant was stillborn.

At least 57 chromosome variants were reported. Inversion 9/9qh+ was by far the most common, occurring in 35 cases. Most of these were in women referred for late maternal age, although two had low maternal serum AFP and six had abnormal ultrasound examinations. These pregnancies usually ended in live births, although two of the AMA women had stillbirths. One other woman with a fetus with 1qh+ also had a stillbirth at 22 weeks' gestation. The other common variants observed were 15p+ seen in three cases and inversion Y seen in five.

Although the number of cases with abnormal results on chromosomal examination is small (in total 399), the proportion of women with abnormal chromosome results who opted for termination of pregnancy was similar both for those having chorionic villus sampling and for those choosing amniocentesis (85% for autosomal trisomies and 75% for sex chromosome anomalies in both).

Recommendation:

Given the relatively high frequency of chromosomal anomalies other than Down syndrome and trisomy 18 detected by invasive testing, all women considering prenatal diagnosis should be informed initially of the possibility of such diagnoses and that, in some cases, parental karyotyping may be required before definitive counselling is possible.

Biochemical and Molecular Diagnoses

The information on outcomes of pregnancies tested for disorders detectable by biochemical analysis or molecular genetics techniques is incomplete, but there were at least three terminations of pregnancy for Duchenne type muscular dystrophy and others of fetuses affected with, or at high risk for, I-cell disease, GM_1 gangliosidosis, myotonic dystrophy, cystic fibrosis, Wiskott-Aldrich syndrome, and thalassaemia. At least two affected pregnancies, one with adrenoleukodystrophy and one at high risk for Becker's muscular dystrophy, were carried to term.

Structural Abnormalities of the Fetus or Infant

It is not possible on a country-wide basis to estimate rates of structural anomalies in the fetuses of referred women because not all centres reported such defects in the outcome data sent to us. This means it is unclear what denominators could be used to calculate rates. However, some information is available.

In total, 62 cases of neural tube defects were identified in the population seen for prenatal diagnosis (1 per 343 total women referred) in provinces other than Manitoba, with two cases detected only at birth. Most (63%) of these cases were detected at ultrasound examination and referred to Genetics for this reason. In Manitoba in 1990, 24 cases were ascertained, 14 having been prenatally diagnosed (1 per 662 women having prenatal diagnosis or screening). Only 21 percent of these were detected through ultrasound examination rather than through routine maternal serum AFP screening.

For Canada as a whole, the cases of neural tube defects can be divided into those ascertained first through abnormal ultrasound findings and those detected by prenatal diagnosis initiated after maternal serum AFP screening, referral for amniocentesis because of late maternal age, or previous history of neural tube defects. Of the 42 cases found unexpectedly at ultrasound examination, there were 26 therapeutic abortions and four stillbirths. The remaining 12 infants were live born and all had spina bifida; eight were from the Maritimes. Among the group of 32 neural tube defects determined prenatally through other indications, all but two pregnancies were terminated. These cases, both in Manitoba, included one stillborn infant with an encephaly whose mother elected to continue until term and one with an anterior meningocele that was corrected after delivery.

There appears little doubt that most women who had a fetus with a neural tube defect prenatally diagnosed elected to terminate the pregnancy. The difference between the group detected through abnormal ultrasounds and the others relates primarily to gestational age at diagnosis. Although anencephaly was detected at similar gestational ages in the two groups, spina bifida was not reported as detected before 20 weeks by ultrasound alone. In Manitoba, most cases of neural tube defects detected by maternal serum AFP screening were diagnosed between 16 and 21 weeks, with a mean of 19 weeks, while those detected by ultrasound alone were picked

up later, between 19 and 32 weeks, with a mean of 25 weeks. Termination of pregnancy was not available for spina bifida in St. John's. However, the gestational ages at diagnosis for the spina bifida cases from that centre ranged from 20 to 30 weeks, with an average of 26 weeks.

Eighteen infants with abdominal wall defects were reported. One was detected at amniocentesis for late maternal age and one by maternal serum AFP. The others were identified initially by ultrasound examination. Eight had omphaloceles, eight had gastroschisis, and the remainder had more severe body wall defects. There were five terminations of pregnancy and two stillbirths among the 15 cases with known outcomes. The others ended in live birth, presumably after amniocentesis had confirmed a normal karyotype. One infant had polydactyly, one had cleft palate, and one had Beckwith-Wiedemann syndrome.

At least three women at risk for recurrence of skeletal dysplasia had an affected fetus detected prenatally. Two women with fetuses with osteogenesis imperfecta and Robert's syndrome respectively terminated their pregnancy. Another woman, carrying a fetus with osteogenesis imperfecta, continued the pregnancy.

Many other cases were referred to the genetics centres because of abnormal ultrasound findings. Over 77 fetuses with major defects were identified. The most common were urinary tract anomalies, central nervous system malformations such as hydrocephaly and holoprosencephaly, cardiac defects, diaphragmatic hernia, hydrops, and cystic hygroma. (Fetuses with cystic hygroma who were found to have chromosomal defects were excluded from this group and included in the cytogenetic anomalies section.) Of the 69 cases in which outcomes are available, only 29 (42%) ended in termination of pregnancy; however, the mortality rate among the others was high, with 40 percent ending in stillbirth or neonatal death. Gestational age at diagnosis was not known for most of these pregnancies; thus, many may not have been detected until late in the second or in the third trimester.

Abnormal Outcomes After Normal Test Results

Outcomes of pregnancies with apparently normal test results are available from seven centres that performed a total of 5 075 amniocenteses and 589 chorionic villus sampling tests. Correcting for pregnancies with incomplete outcome data and those with abnormal results, approximately 4 514 amniocenteses and 438 chorionic villus sampling tests had apparently normal results. These centres reported 29 spontaneous abortions after amniocentesis (0.6%) and six (1.4%) after chorionic villus sampling. Three additional women had therapeutic abortions for unrelated reasons. After amniocentesis, an additional 20 women had stillbirths (0.4%) and there were two neonatal deaths. No perinatal deaths after chorionic villus sampling were reported. These data must be interpreted with caution; both the denominators and numerators of these rates are potentially inaccurate, and the ascertainment of abnormal results after

amniocentesis and chorionic villus sampling may have varied in completeness. In addition, some of these women may have had medical conditions affecting the likelihood of these outcomes, but we did not collect data on the health status of the women themselves.

A variety of congenital malformations were reported in infants born after prenatal testing. Although most were minor, such as hydrocele, pilonidal dimple, haemangioma, and club foot, other abnormalities such as cleft lip, cleft palate, cardiac defects, and tracheoesophageal fistula also occurred. Among the chorionic villus sampling patients, one was reported to have had a child with limb malformations and further information is being sought. The possibility of an increased incidence of limb deficiency defects among infants subjected to chorionic villus sampling has been raised, but the issue is still controversial (Firth et al. 1991a, 1991b; Froster and Baird 1992; Hsieh et al. 1991; Jackson et al. 1991; Mahoney 1991; Mastroiacovo and Cavalcanti 1991; Miny et al. 1991; Monni et al. 1991; Schloo et al. 1992). Most of these defects have occurred after very early chorionic villus sampling, usually before 10 weeks' gestation. Further detailed epidemiological evaluation of the data worldwide is being attempted.

Recommendation:

In view of the possibility that there is an increased risk of limb deficiency defects after chorionic villus sampling, particularly when performed very early in gestation, we recommend that the procedure not be performed before 10 weeks' gestation until definitive epidemiological information is available.

False Negative and False Positive Results

Very few true false positive or false negative results were reported by centres. There were three reports of inaccurate sex determination, including both an apparently normal girl after a 46,XY result and the converse. The reasons for errors include contamination of the sample by maternal cells, an undiagnosed twin, sexual differentiation disorders, and human error. Such errors are apparently rare. In one other case, the sex of the infant was incorrectly given over the telephone, although the test result indicated the appropriate sex.

In one case, a polyploid 92,XXXX karyotype was reported after a chorionic villus sampling was performed at 17 weeks' gestation. The scheduled amniocentesis had been deferred because of oligohydramnios. At 19 weeks an amniocentesis was done to re-evaluate the first result, but the patient elected to terminate the pregnancy before the results (normal) of the amniocentesis were available.

The survey design does not allow estimation of the frequency of false negative and false positive ultrasound diagnoses. However, in at least three cases the finding of choroid plexus cysts on ultrasound led to amniocentesis with normal results. The significance of such cysts as an indication for invasive testing is also being evaluated. They are a relatively common finding in second trimester ultrasonographic examinations but are potentially associated with an increased risk for aneuploidy (Achiron et al. 1991; Chinn et al. 1991; Platt et al. 1991; Porto et al. 1992; Twining et al. 1991).

Decision-Making Processes

Centres were asked about awareness of the Canadian guidelines for the delivery of prenatal diagnostic services; all indicated they were aware of these. Most of the centres indicated that they had a committee, either within the unit and comprising members of the unit, or less frequently including representatives from other disciplines. One centre indicated that this committee included a representative from the chaplaincy office of the institution. None of the committees included lay or consumer representatives. One large centre had developed an extensive committee structure to deal with prenatal issues:

- 1. prenatal working group with representatives from both genetics and obstetrics departments (this was primarily a problem-solving and development group);
- 2. advanced maternal age working group to resolve issues relating to two hospitals where the testing was done;
- 3. Joint Prenatal Diagnosis Committee (sets policy for the prenatal program);
- 4. Provincial Genetics Advisory Committee (representatives from genetics department, hospital administration, and provincial government).

However, this structure appears to have no formal mechanism for the incorporation of views of consumers and women's groups.

Recommendation:

It is our view that all centres should formalize an appropriate committee structure to ensure that prenatal policy is regularly and adequately discussed. This committee should include not only caregivers, but also someone with some knowledge of ethical principles and representatives of consumer and women's groups. If it is not thought appropriate to include consumer groups on the committee, arrangements should be made at least annually to meet with such groups to learn of their concerns.

Staffing

Number of Staff

The data provided by the centres on staffing for prenatal diagnosis were extremely variable in quality. Since most centres provided the number of physicians and others involved in prenatal diagnosis, all data were converted to the number of people available to see patients for prenatal counselling, recognizing that most, if not all, have other duties as well. To try to make comparisons between provinces in terms of possible work load, we have taken as the target population for prenatal counselling in 1990 the number of women who were over the age of 35 and who gave birth in 1989 (latest Statistics Canada data available).

Table 26 shows the total number of people available to provide prenatal counselling, by their primary qualification. Table 27 gives the ratios of available staff to numbers of mothers over 35 years of age. In 1990, in Canada as a whole, there was one M.D. or Ph.D. available for genetic counselling of prenatal cases per 474 women over 34 years of age, and one genetic counsellor per 565 women over 34. Examination of these data province by province showed considerable variability, from 1 in 925 in Quebec to 1 in 222 in Manitoba, for M.D.s and Ph.D.s. The ratio of genetic counsellors ranged from 1:217 in Newfoundland to 1:1 296 in Quebec.

The variation between provinces reflects several factors, including the budget available and mode of payment for services, priorities given to development of prenatal services, and patterns of practice in individual provinces. For instance, in Manitoba most prenatal counselling was done on an individual basis by either an M.D. or a Ph.D. genetic counsellor, and very few, if any, patients were counselled by their obstetrician or family practitioner. In British Columbia, most patients who were of advanced maternal age were counselled by their physician in the community, and came into the centre simply to have the fluid drawn. Chorionic villus sampling patients were counselled at the time the procedure was done. In Quebec, only chorionic villus sampling patients were counselled in the genetics centres. Most women having AMA amniocenteses were both counselled, and had the fluid drawn, by the community obstetricians.

The data from Alberta do not include staffing of the outreach units. In Edmonton, patients were counselled by outreach nurses and fluids were drawn in the community and sent to the laboratory. In the Northwest Territories, amniocentesis was available in Yellowknife and samples were processed in Edmonton. In Saskatchewan, access to services depended largely on residence. In Saskatoon or areas served by the Saskatoon centre, most patients were seen by a geneticist or genetic counsellor; in Regina and southern Saskatchewan, patients were counselled in the community. Of all the provinces, prenatal diagnosis services in Saskatchewan were among the least developed, with only 22.8 percent of mothers over 35 years of age receiving testing. Only Newfoundland was

Table 26. Personnel Involved in Prenatal Counselling, by Region (Does Not Include Outreach)

Personnel involved in prenatal counselling through genetics	British Columbia	Alberta	Saskatchewan	Manitoba
No. of M.D.s	5*	7	2*	4
No. of Ph.D. counsellors [†]		1		2
No. of genetic counsellors ^{††}	12	7	. 2	3 [‡]
Total	17	15	4	9

^{*} There is also one other CCMG-accredited physician in private practice in Victoria and one in Saskatoon.

Table 27. Availability of Medical Geneticists and Genetic Counsellors

					-
	British Columbia	Alberta	Saskatchewan	Manitoba	Ontario
No. of women over 35 years of age	4 319	3 278	891	1 334	20 420
No. of M.D.s, Ph.D.s	5	8	2	6	32
No. of M.D.s, Ph.D.s/1 000 women over 35	1.15	2.44	2.24	4.5	1.57
No. of genetic counsellors	12	7	2	3	25
No. of genetic counsellors/ 1 000 women over 35	2.78	2.13	2.24	2.25	1.22

^{*} Excludes Ph.D. laboratory scientists.

^{**} No information from Hamilton.

^{***} One of the CCMG-accredited physicians in this centre does minimal counselling.

Ontario					
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
18	10**	7	6***	2	61
3	1			1	8
16	9	. 5	1	2	57
37	20	12	7	5	126

See Table 29.

See Table 31.

Includes the maternal serum AFP program coordinator (an interdisciplinary provincial program).

Quebec	Maritimes	Newfoundland	Northwest Territories	Total
6 481	1 431	434	131	38 719
7	6	3	0	69*
,	0	3	O	69
1.08	4.2	6.9	0	1.78
5	1	2	0	57
0.77	0.69	4.6	0.00	0.69

lower at 15 percent. Quebec, with the lowest ratio of geneticists and genetic counsellors, had the highest proportion of services (64.5%) (see section entitled "Proportions of Eligible Women Referred for Prenatal Diagnosis").

Qualifications of Staff

Of the 61 M.D.s involved in prenatal diagnostic services in the Genetics centres (Table 26), 40 (66%) were accredited by the CCMG (Table 28). Of those, 34 were accredited in clinical genetics, three in cytogenetics, two in molecular genetics, and four in biochemical genetics. Three M.D.s were accredited in more than one specialty. Of the 41 Ph.D.s involved in prenatal diagnosis (Table 29), 33 (80%) were involved in one or more of the laboratory disciplines, 13 in cytogenetics, 10 in molecular genetics, and five in biochemical genetics. Five Ph.D.s were involved in maternal serum AFP analysis and interpretation. Eight Ph.D.s were specifically involved in prenatal counselling (CCMG accreditation of Ph.D.s is given in Table 30). Of the 21 accredited Ph.D.s, 16 (76%) were accredited in a laboratory discipline and only five (24%) in medical genetics. Ph.D.s were involved in some aspect of service delivery in all regions of Canada except British Columbia.

Fifty-seven genetic counsellors were involved in delivery of prenatal diagnostic services across Canada (Table 31). Of these, 14 (25%) had formal training in genetic counselling; four had a master's degree in genetics; and 32 were trained as nurses, with 50 percent having a nursing degree and 50 percent a diploma. Seven had some other form of training. Only McGill has a genetic counsellor training program that leads to a master's degree. Thus, many of the formally trained genetic counsellors would have received their training in the United States. Duties performed by genetic counsellors varied between centres; these ranged from clinic coordination with minimal involvement with counselling, to extensive involvement with counselling of patients with complex genetic problems where the diagnosis had been confirmed by a geneticist/M.D. In these cases a detailed family history was taken and the options available were explained so that the woman or couple could make an informed choice.

Recommendations:

Adequate fellowship training support should be provided by the federal and provincial governments to provide training for accreditation by the CCMG for Ph.D.s wanting to work in genetic service laboratories.

Universities should consider development of suitable interdisciplinary training programs at the master's level for individuals wishing to undertake careers as genetic counsellors.

(cont'd)

The CCMG and the CAGC should be encouraged to include genetic counsellor training programs in the CCMG accreditation of centres program.

The CAGC should be encouraged to develop an accreditation program for Canadian genetic counsellors, and each centre should be encouraged to develop an appropriate career structure for such individuals, if not already done, either through an affiliated university or hospital or through their provincial program, whichever is the most appropriate.

Genetics centres should be encouraged to review their programs to ensure that appropriately trained staff are filling positions and, when hiring new personnel, that the most cost-effective solution is followed.

Staffing Requirements by 1995

Centres were asked to predict the number of additional staff they thought would be required by 1995 compared to the staff they had in 1990. The responses to this question are given in Table 32 for prenatal counselling and Table 33 for laboratory personnel. Significant increases were predicted for M.D.s, genetic counsellors, and outreach personnel. Increases were also predicted for laboratory personnel with an additional 36 full-time equivalents being required at the M.D./Ph.D. level, and 170 more full-time equivalent technologists, of whom 70 percent would be required in cytogenetics laboratories. Predicting staffing needs in a period of service expansion on the one hand and fiscal retrenchment on the other is at best providing an informed guess as to the real needs of the service in the future. It is unclear to us how much of this expanded staffing requirement was realistic and how much was a wish list. However, it seems that much of the service expansion in prenatal diagnosis will come from increased use by the AMA group (this may plateau with time). Another factor is increased fertility in this age group, with delayed childbearing increasingly common in Canada. Also, improved maternal serum screening for chromosome abnormalities among women in the 30to 35-year-old group will increase the numbers found to be at increased risk of a fetus with chromosome abnormality. In addition, there will be a significant expansion in the number of molecular prenatal diagnoses requested. However, as the technology improves, the time needed for and the complexity of these tests will likely be reduced.

We would question the prediction that a large increase in the number of physicians will be required for prenatal counselling. Because of the nature of the profession, billing, and fee structures, physicians performing a service for which they are not strictly required generally cost more than

Table 28. CCMG Sub-Specialization of M.D.s Involved in Prenatal Diagnosis in Genetics Centres, by Region

	British Columbia	Alberta	Saskatchewan	Manitoba
Total no. of M.D.s with CCMG accreditation	5**	7	1**	4
No. accredited in clinical genetics	5	6	1	4
No. accredited in cytogenetics				
No. accredited in molecular genetics	1			
No. accredited in biochemical genetics		1		
No. accredited in more than one sub-speciality	1			

^{*} No information is available from Medical Genetics in Hamilton.

Table 29. Ph.D. Involvement in Prenatal Diagnosis, by Region

No. of Ph.D.s involved in	Alberta	Saskatchewan	Manitoba
Prenatal counselling	1		2
Cytogenetics	3	1*	1
Molecular diagnosis	2		
Biochemical diagnosis	1		
Maternal serum AFP determination	1		
Total	8	1*	3

^{*} This individual is a D.M.V.

Note: Some Ph.D.s fit into more than one category (i.e., there is only one Ph.D. involved in the prenatal program in Newfoundland; this person is involved in cytogenetics and does some counselling). Vancouver, McGill, Credit Valley, North York, Victoria, and Regina have no Ph.D.s involved in prenatal diagnosis.

^{**} There is another CCMG-accredited physician in private practice in Saskatoon and one in Victoria.

Ontario					
Toronto	Excluding Toronto*	Quebec	Maritimes	Newfoundland	Total
11	4	5	2	1	40
9	4	3	1	1	34
1		2			3
		1			2
0			4		4
2					4
1		1			3

	Ontario				
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
3	1			1	8
2	4		1	1	13
1	3	3	1		10
1	1	1	1		5
1	2	1			5
8	11	5	3	2	41

Table 30. CCMG Sub-Specialization of Ph.D.s Involved in Prenatal Diagnosis, by Region

				Ontario			
CCMG accreditation	Alberta	Manitoba	Toronto	Alberta Manitoba Toronto Excluding Toronto Quebec Newfoundland Total	Quebec	Newfoundland	Total
Medical genetics	-	2	2				2
Cytogenetics	2	-	7	4		-	10
Molecular genetics	-		-	-			က
Biochemical genetics	-			1	-		m
Total	5	ო	22	9		-	21

Note: Some Ph.D.s are accredited in more than one CCMG sub-speciality. Vancouver, McGill, Credit Valley, North York, Victoria, and Regina have no Ph.D.s involved in prenatal diagnosis.

non-physicians for the same service. Therefore, we do not see the need to have M.D.s running laboratories (except in unusual circumstances) or counselling patients of AMA in straightforward cases. We believe that much of the AMA counselling could be done by trained genetic counsellors, and that women may be more likely to be appropriately counselled by them than by their family physician or obstetrician, who may not have adequate knowledge or the time to discuss the various options available.

Budget and Remuneration

Centres were asked to provide data on the costs of the prenatal service, separated as far as possible into the components of genetic counselling, laboratory costs, and procedure costs. Data received from the centres were extremely variable, with some centres providing unit costs and others providing total amounts budgeted for the particular procedure. When centres presented global budgets, estimates were made of costs per procedure by dividing the total budget by the number of procedures.

Centres were also asked to specify the mode of remuneration for personnel, and to indicate whether they thought the method used was appropriate given their particular circumstances. Because health care costs in Canada are paid by provincial medicare programs, costs for a given service within an individual province should be approximately equal between centres within the province.

The data in Table 34 should be taken as estimates based on the figures provided by each centre. Data are presented only from the centres providing minimal data for analysis. In very rough terms the cost of an amniocentesis in Canada, including counselling session, laboratory costs, procedure costs, and at least one billed ultrasound, ranged from \$430 in Ottawa to \$2 020 in Newfoundland. The high cost of the service in St. John's represents the necessary costs required to maintain the service, and the fact that a relatively small number of procedures are done in that province. The cost of a chorionic villus sampling procedure and related tests and laboratory procedures ranged from \$460 in Ottawa and Quebec to \$925 in London. In most centres there was little difference in cost between an amniocentesis and a chorionic villus sampling despite the increased work load in the laboratory and the increased skill required by the operator. Significant variation in costs appeared to occur between different centres in Ontario despite the fact that all were funded by the Ontario Ministry of Health by means of global budgets. These differences may represent different allocations to centres depending on case load and types of laboratory services provided.

In three centres, M.D.s were remunerated by salary for provision of prenatal diagnostic services; all considered this to be appropriate. Four centres remunerated their M.D.s on the basis of fee for service alone; two of the four considered this mode of remuneration inappropriate and two

Table 31. Number and Training (Highest Degree Obtained) of Genetic Counsellors Involved in Prenatal Diagnosis, by Region (Not Including Outreach)

<u> </u>	British Columbia	Alberta	Saskatchewan	Manitoba
Formal training in genetic counselling	6	1		1*
Master's degree in genetics	2			
Nursing degree	2	3	1	
R.N. diploma	2	1	1	2
Other		2		
Total no. of genetic counsellors	12	7	2	3

Maternal serum AFP screening program coordinator.

Table 32. Anticipated Additional Full-Time Equivalent Staff Required for Prenatal Counselling by 1995, by Region (%)

	British Columbia	Alberta	Saskatchewan	Manitoba
M.D.s	4.0	2.0	2.0	1.5
Ph.D.s		1.0	1.0	0.5
Genetic counsellors	6.0	3.0	1.0	1.5
Nurses/R.N.s		2.0		
Outreach personnel	3.0	6.0	3.0	1.0
Support staff	14.0	4.0	1.0	2.5

considered it appropriate. Three centres remunerated by both fee for service and a salary; all considered this appropriate. In all centres, Ph.D.s and others providing service were remunerated by salaries and they felt this to be appropriate. One centre commented that salaries were an appropriate way to remunerate clinical geneticists and others counselling prenatal patients, since it removed any conflict-of-interest possibilities.

	Ontario				
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
			*		
3	2	1		5	14
1		1			4
6	2	1		1	16
5	2	2	1		16
1	3			1	7
16	9	5	1	2	57

	Ontario				
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
9.0	8.2	6.0	6.0	1.0	39.7
1.0	2.0				5.5
11.2	7.5	5.0	3.0	2.0	40.2
2.9	1.0	3.0			8.9
1.5	4.0		1.0		19.5
6.5	8.5	4.0	1.0	1.0	42.5

Legal and Ethical Problems

Confidentiality was raised as an issue by one centre where a limited number of cases was seen. Three other centres raised the issue of confidentiality with respect to third parties, when testing inadvertently

Table 33. Anticipated Additional Laboratory Staff (Full-Time Equivalent) Required by 1995 for Prenatal Diagnosis, by Region (%)

	British Columbia	Alberta	Saskatchewan	Manitoba
Cytogenetics laboratory personnel				
M.D./Ph.D. cytogeneticists	3.0	2.0	1.0	1.0
Technologists	10.0	11.5	3.0	7.0
Molecular/biochemical laboratory personnel				
M.D.s/Ph.D.s	3.0	3.0	1.0	1.0
Technologists	7.0	6.0	1.0	2.0

Table 34. Proce	edure Costs (\$	5)			
Service	Vancouver	Edmonton	Winnipeg	Ottawa	Kingston
Genetic counselling		75	74.50	106	100
Laboratory costs	550	195	285	166	480
Procedure costs					
Amniocentesis Chorionic villus	125	110	317	51	
sampling	150	144	317	80	
Ultrasound	80	116	75	105	
Shipping	30	3			
Salaries	yes	yes	fee for service	yes	
Amniocentesis					
total		496	677	430	645
Chorionic villus		F20	677	400	0.45
sampling total		530	677	460	645

TGH — Toronto General Hospital; HSC — The Hospital for Sick Children; OHIP — Ontaric Health Insurance Plan.

	Ontario				
Toronto	Excluding Toronto	Quebec	Maritimes	Newfoundland	Total
2.4	4.0	3.0	1.0	1.0	18.4
19.5	36.0	22.0	9.0	1.0	119.0
0.8	6.0	2.0	2.0		18.8
4.1	14.0	4.0	13.0		51.1

London	TGH	HSC	North York	Halifax	Quebec	St. John's
96			132	60	37	663
425 AM 670 CV	333 368	368	648	307	325	1 176
52	OHIP	OHIP	108		40	52
83 76	52		91	110	50	139
alary & fee for service	fee for service	salary & fee for service	fee for service	salary & fee for service		fee for service
649			979		450	2 020
925			n.a.		460	

demonstrated that other family members were at risk, but the individual first seeking genetic advice refused consent to disclose. This is a general ethical issue that arises sometimes in medical genetics practice and is not confined to prenatal diagnosis.

Several centres raised the issue of recurrent requests for prenatal testing when no other medical indication exists, to determine the sex of the fetus and abort if not the sex of choice. Although this issue was raised as a concern by several centres and may involve significant counselling time, it seems to be an extremely rare occurrence in Canada (see section entitled "Non-Medical Reasons for Requests for Testing"). Centres documented only 14 cases in 1990 and in only one was invasive testing performed. Despite the findings of Roy and Hall (1989) that 30 percent of their respondents would do such testing, it seems to occur seldom and is strongly discouraged by centres. A more complex issue was sex selection in patients with valid medical indications for prenatal diagnosis such as advanced maternal age. Some patients eligible for the test for other reasons may disclose at the time of counselling that a fetus with normal chromosomes. but not of the sex of choice, will be aborted. It is our view that centres are not justified in withholding information about the sex of the fetus because the patient has disclosed her intentions at the time of testing. In our view, centres are justified in refusing to facilitate the termination of a normal pregnancy for the reason of sex alone.

Late termination of pregnancy and possible fetal viability was raised by one centre (Ste. Justine). Another (St. John's) raised its concern about lack of access to second trimester terminations of pregnancy for genetic reasons other than anencephaly. The section entitled "Decision-Making Processes" refers to the decision-making processes in the centres, and we emphasize the importance of establishing a formal committee structure in each that would meet at least annually to establish policies and deal with any complex legal or ethical issues.

Discussion

In many respects, Canada has provided leadership in the areas of safety and regulation of prenatal diagnosis for genetic disease. The country was the first to develop a national set of guidelines concerning the use of such techniques and pioneered the first randomized controlled trial of chorionic villus sampling. The establishment of the CCMG was another example of this trend and acknowledged the concern the medical geneticists had that genetic services must be delivered in an effective, safe, and non-directive fashion. With time it has become apparent that a need exists for more education of other health care professionals and the public concerning the implications of genetic testing and more involvement of the consumers — the families — in decisions about what services should be

made available and their order of priority. The establishment of the Royal Commission on New Reproductive Technologies is a reflection of this increasing awareness.

The undertaking of this research project was to describe as accurately as possible the current status of prenatal diagnosis of genetic disease in Canada. One objective was obviously to document whether concerns expressed by the public in the forum of open Commission hearings did reflect valid understanding of current practice. Two major topics were raised on several occasions: that prenatal diagnosis was being used for sex selection and that women were being coerced into agreeing to terminate affected pregnancies as a precondition of having prenatal testing. Although these subjects were discussed in depth with the centres providing these services across the country, little evidence was found to support such allegations. Centres reported that a few women requested sex selection on non-medical grounds but, with one exception, such requests were denied. Respondents from centres universally found this idea repugnant and sometimes were concerned because tests could be done for valid medical reasons but the results could be used in this way. In no centre was agreement to terminate an affected pregnancy a prerequisite for testing, although one centre admitted it discouraged testing where a woman was adamant that she would not consider this option. We believe a very clear distinction should be made between the testing process and the decisions to be made concerning abnormal results.

It is impossible to ignore the strong feelings some people have that prenatal testing, with its ability to identify individuals with genetic disorders, may lead to a devaluing of the human life and potential of such infants. However, it is also impossible to disregard the fact that genetic disorders are becoming increasingly important as contributors to childhood mortality and morbidity and predisposing factors for adult onset disease. The burden of these conditions is high in economic terms, but even more pressing with respect to the social and emotional costs borne by individual patients and their families. The fact that over 22 000 women in Canada used prenatal genetic services in 1990 indicates an interest in, and awareness of, these programs. What is less clear is the extent to which women in different parts of the country are being informed about genetic testing, the type of information they receive, and the ease with which they can gain access to the types of testing or counselling and information they want.

Prenatal genetic services tend to be concentrated in the major university teaching hospitals or large community hospitals. This may have had the unfortunate consequence of "medicalizing" the approach to provision of such services and affecting the usually optimistic time of pregnancy by introducing unnecessary anxiety. On the other hand, most women have concerns that their baby may not be healthy, and this apprehension is greater for women at increased risk due to their age or family history. A woman's desire to avert potentially avoidable, unfortunate outcomes of pregnancy would seem to make the rise of the "tentative

pregnancy" inevitable, despite the potential problems engendered by prenatal testing (Tymstra 1991). One of the objectives of prenatal diagnostic services is, therefore, the provision of information concerning risks and options in an accurate, unbiased, and reassuring manner.

It seems logical that women who receive information about prenatal diagnosis in the context of their general medical care and by their local health care providers would have less anxiety provoked by the process and feel more comfortable in making their own decisions whether or not to have prenatal diagnosis than those counselled in more formal settings. Thus, the availability of appropriate written material and personal counselling at the local level would seem to be necessary. This need has been recognized in several provinces that have developed effective outreach programs and would seem to work well where the medical geneticists work closely with the public health nurses and community physicians. However, there is a potential danger that women counselled in the community may be misinformed about the types and risks of testing or are unduly pressured by well-meaning but biased people to be tested or to forgo testing. More research is required at the community level to determine the quality and quantity of information being provided and the methods of counselling that are most acceptable to the women concerned.

The most common reason for referral for prenatal diagnosis in 1990 was late maternal age (35 years of age or older). In Canada, about 52 percent of such women were referred to genetics centres for counselling or testing or both. However, the proportion of eligible women referred varied considerably from about 65 percent in Quebec to 15 percent in Newfoundland. The reasons for such differences are probably also variable. In Quebec, many amniocenteses are done in local hospitals and the fluids sent to centre laboratories for analysis; thus, women do not have to travel large distances to be tested. However, distance alone or the presence of outreach services would not appear to be the only factor involved. In Manitoba, almost without exception, women who want counselling and invasive testing must travel to Winnipeg; yet the proportion of eligible women referred is relatively high at 49 percent compared to Alberta (30%). which has an extensive outreach program. Other factors influencing the acceptance of referral by women in different areas must include the likelihood of being informed about the availability of testing and their willingness to participate. Both physicians' and patients' attitudes and perceptions are obviously important, but a general lack of local medical services may be more so. We note with interest the preliminary data from MacLeod et al. (1993) that the distribution of physicians and distance from genetics centres appear to influence frequency of referral in the Maritimes and we look forward to the results of their studies in other regions. We also are concerned with the results of Chodirker and Evans' (1993) study that rural physicians in Manitoba appear less well informed concerning indications for prenatal diagnostic referrals or the implications of abnormal maternal serum AFP results.

Whatever the reasons for differences in uptake of services for late maternal age, they appear to have a similar impact on referral for other reasons. Women in Newfoundland, the Maritime provinces, and Saskatchewan may be less interested in being referred for genetic testing, but it seems unlikely that this factor alone is responsible for only 1.5 percent to 3 percent of pregnant women in these populations being referred compared to 6 percent in Quebec, Ontario, Manitoba, and British Columbia.

In general, the prenatal diagnostic services available to women in 1990 in Canada included genetic counselling, invasive testing such as amniocentesis and chorionic villus sampling, and non-invasive tests including ultrasonographic examination and maternal serum AFP screening. Although all centres followed the Canadian guidelines concerning the indications for prenatal testing, the actual details of services available and the specific mechanisms used to provide them varied across the country.

Counselling of families concerning the risks and options involved was seen as a major component of prenatal diagnosis programs but, in many regions, the responsibility for this activity rested with the primary care physician. We have little information on the type of counselling provided, as few centres had any feedback from the physician concerning family history or other pertinent factors. Where patients tended to be referred to the genetics centre for counselling, appropriately trained genetic counsellors often acted as a major resource for counselling patients at many stages of the testing process. They often took family histories and counselled families about the tests available and were involved in the ongoing support of families with abnormal results. Presumably, such people have chosen a career that allows development of specific expertise in genetic counselling and fulfils their desire to work with families; thus, it would seem appropriate that they be encouraged to take a more active role in prenatal counselling and patient support in centres where these are not their presently defined primary roles.

Most women seen in the genetics centres subsequently have an invasive procedure. This is not unexpected because most referrals for testing were because of late maternal age and increased risk of chromosomal abnormalities. Amniocentesis was available to women throughout the country, although many had to travel considerable distances to be tested. However, chorionic villus sampling was an option for only some Canadian women. A direct relationship seems to exist between the availability of chorionic villus sampling (expressed as numbers of tests performed) and the proportion of eligible women so tested. This was not seen for amniocentesis. In particular, women who had previously had a child with a genetic disorder or were at high risk were more likely to opt for the earlier test where it was available. Chorionic villus sampling seems to be a test of choice for many women electing to have prenatal diagnosis; however, their access to it is obviously directly influenced by its

availability. Even in provinces where the test was provided, some women may have been referred too late for it to be an option. A study in the Netherlands identified late referral as the major factor restricting uptake of chorionic villus sampling and noted that women having this procedure were informed about prenatal testing earlier in their pregnancies than those having amniocentesis (Brandenburg et al. 1991).

Despite the desire of some eligible women to have chorionic villus sampling, the test has drawbacks. In particular, the need in some cases for repeat testing is problematic. This is related to the number of such tests performed, probably due to the need to do a certain volume to develop and maintain the skill needed. For this reason, it would seem appropriate to recommend that, at this time, chorionic villus sampling be done by a limited number of obstetricians and in a relatively small number of centres. Women wishing the earlier test, especially those at high risk, should be referred to the nearest centre performing the test and, where necessary, the financial burden for this should be borne by their provincial health care plan. At the same time, the planned research into the feasibility of early amniocentesis should be supported because such a form of testing is more likely to become readily available locally.

With respect to invasive testing and coercion, it should be noted that a significant number of eligible women who were referred in fact declined the procedures offered. About 1 in 10 women decided after counselling not to have invasive testing. It is of vital importance that counselling be non-directive and that women be given the opportunity to receive accurate information about testing and maintain the freedom to make their own decisions in light of that.

Invasive testing, by its nature, requires the support and cooperation of the genetics centres because few amniocenteses are performed in the community and samples are referred to private laboratories. With the non-invasive tests, such as ultrasound and maternal serum AFP, the influence of the medical genetics community on their use for prenatal diagnosis is clearly much weaker. The relatively routine use of ultrasound may have the unavoidable consequence of it becoming a screening tool for fetal anomalies. Unfortunately, the detection of such anomalies often occurs late in gestation when termination of pregnancy is no longer an option. For some abnormalities, especially neural tube defects, maternal serum AFP offers the opportunity for earlier diagnosis and may have added benefits with respect to identification of other high-risk pregnancy situations. However, maternal serum AFP screening also has problems. Quite rightly, genetics centres in many provinces have been cautious in proceeding with screening until the appropriate feasibility studies have been done, priorities assessed, and resources made available; however, this reticence on the part of genetics centres has not stopped the introduction of screening in many areas, and women are likely being screened without appropriate information being provided and without adequate care being taken to ensure accurate interpretation and follow-up. Provincial health

care programs should review the extent of community screening in their areas and ensure that the established guidelines for maternal serum AFP

screening are being followed.

The three most widespread tests performed in Canada for genetic prenatal diagnosis are amniocentesis and chorionic villus sampling, for women of advanced maternal age, and maternal serum AFP testing. One or other of these could have an impact on every pregnancy. However, major discrepancies are obvious when we compare the care taken to ensure that these tests are available to women who want them to the options offered to the very small number of patients requiring diagnosis for a biochemical or metabolic problem. There is an excellent network of laboratories performing molecular and biochemical tests with well-established cooperation between the provinces. It seems ironic that the genetic services supplied to patients requiring uncommon molecular and biochemical diagnoses in many provinces are far superior to those available to the much larger population of pregnant women in Canada who will have most infants with potentially preventable genetic disease. With the continued development of screening tests such as maternal serum AFP and triple testing, which will be pertinent to all pregnant women, re-evaluation of the resources available for prenatal testing will have to be made. Obviously, families that have already had a child with a genetic disorder must have the opportunity for counselling and prenatal diagnosis, if it is feasible for the condition. A different kind of commitment is needed to offer choice for the more common situations.

The end-point for any pregnancy comes with spontaneous or therapeutic abortion, stillbirth, or live birth. It would appear necessary in the evaluation of any prenatal program that information on pregnancy outcomes should be available. Most centres doing prenatal diagnosis had mechanisms to collect such data, but some did not. Others followed up women with abnormal results to confirm the diagnosis, but did not trace those with normal findings. It would seem appropriate that follow-ups are done so that the accuracy and safety of procedures being offered can be

monitored by every program.

For women who receive abnormal results, the options are obviously limited. Few of the disorders detected are amenable to treatment and most lead to serious physical and mental handicap. In 1990, most of these women elected to terminate pregnancies when an abnormality was detected; however, many did not. Presumably, women were able to maintain their autonomy and make their own decisions in these difficult situations. However, the decisions they made were not arbitrary, perhaps due to the post-diagnosis counselling that was offered. Termination of pregnancy was less common in situations such as sex chromosome anomalies where resulting handicaps may be minimal or in cases of abdominal wall defects where corrective surgery is a possibility. It was not possible to ascertain from the outcome data provided whether the proportion of women electing termination differed depending on whether

they were counselled after diagnosis by a geneticist or a physician in the community. Certainly, from other studies it has been noted that, with sex chromosome anomalies, patients are more likely to continue the pregnancy if counselled by a geneticist (Holmes-Seidle et al. 1987; Robinson et al. 1989; Verp et al. 1988).

Even when the prognosis is grave some women elect to continue their pregnancies, and early diagnosis in these cases may help the family prepare and develop social and economic support systems. For women who find that continuation of the pregnancy is not a situation they can accept, termination should be an option. Unfortunately, in some parts of the country referral for such a procedure may cause inconvenience and economic hardship for a family already experiencing the grief that accompanies an abnormal result.

Despite the sensitive nature of the prenatal diagnostic process and the considerable ethical dilemmas that the new reproductive techniques in this area may present, the genetics centres involved appear to be using these tests responsibly and conscientiously. The centres are conscious of the potential misuse of testing, especially with respect to sex selection, and exercise strong control over the availability of testing for such reasons. However, we would be naive to think that the desire for such testing does not continue among certain pregnant women and that some community physicians, both in Canada and the United States, appear willing to meet their requests.

Prenatal diagnosis of genetic disorders is a widespread and growing field of medical activity, and one that impinges on the lives of all Canadians. It is vital that improved methods of communication be developed between medical geneticists, community health care providers, pregnant women and their partners, and provincial and federal health care funding agencies if such programs are to continue to develop appropriately to meet the needs of Canadian women in the future.

Appendix 1. Site Visit Questionnaire

ASSESSMENT OF PRENATAL SERVICES OFFERED IN A GENETIC SERVICE SETTING

CENTRE	CODE:

A. BACKGROUND INFORMATION

1.	In 1990, was the service component of this centre accredited by the
	Canadian College of Medical Geneticists (CCMG)?

 	Yes/No (Y/N) []

	When did it first become accredited?
	Date of last accreditation:
	If not CCMG accredited, name of accrediting body:
In ce	1990, were the cytogenetic laboratory facilities part of the genetic ntre?
	If separate, to whom did they report?
	Was the director CCMG accredited? Yes/No (Y/N) [] Other affiliation:
	What was the relationship of the laboratory to the genetic centre?
al	1990, were the laboratory facilities evaluating maternal serum pha fetoprotein (MS-AFP) part of the genetic centre? Yes/No (Y/N) [] Was the director CCMG accredited? Yes/No (Y/N) []
	Other affiliation:
_	Other affiliation: What was the relationship of the laboratory to the genetic centre?
	What was the relationship of the laboratory to the genetic centre?
d Ir	What was the relationship of the laboratory to the genetic centre? 1990, did you have laboratory facilities doing molecular prenatal iagnosis? Yes/No (Y/N) []
d Ir	What was the relationship of the laboratory to the genetic centre? 1990, did you have laboratory facilities doing molecular prenatal iagnosis?
d Ir	What was the relationship of the laboratory to the genetic centre? 1990, did you have laboratory facilities doing molecular prenatal iagnosis? Yes/No (Y/N) [] 1990, were the laboratory facilities doing molecular prenatal iagnosis part of the genetic centre? Yes/No (Y/N) []

	1990, did you have laboratory facilities doing biochemical prenat gnosis?
In dia	1990, were the laboratory facilities doing biochemical prenat gnosis part of the genetic centre? Yes/No (Y/N) [
I	f separate, to whom did they report?
Ţ	Was the director CCMG accredited? Yes/No (Y/N) [
	Other affiliation:
Wh	at was the relationship between the genetic centre and the lity(s) that offered diagnostic ultrasound (ie: level II or
Wh	
Wh factults	ility(s) that offered diagnostic ultrasound (ie: level II or rasound)?
Wh fact ultr	ility(s) that offered diagnostic ultrasound (ie: level II or
Who fact ultransport with the work of the	dity(s) that offered diagnostic ultrasound (ie: level II or rasound)? The patient records/information computerized in 1990?

What was the nature of this interaction ie: to formulate/discuss changes in the provision of services, sending out samples when the demand exceeded local capacity, etc.?
Please comment:
In 1990, how many individuals practised Obstetrics in your catchment area ? (Please do not include residents in Obstetrics in these figures.):
<u>n =</u>
Obstetricians
Non-Obstetrician physicians
In 1990, were there individuals independent of the Genetics centre who counselled women and performed amniocentesis? (Please do not include residents in Obstetrics in these figures.)
Obstetricians
Non-Obstetrician physicians
In 1990, did your genetics centre designate specific obstetricians to perform amniocentesis and CVS? Yes/No (Y/N) []
In 1990, how many individuals associated with your centre performed genetic amniocentesis? (Please do not include residents in Obstetrics in these figures.):
Obstetricians
Non-Obstetrician physicians
In 1990, how many individuals associated with your centre performed CVS? (Please do not include residents in Obstetrics in these figures.):
Obstetricians
Non-Obstetrician physicians

17.	In 1990, how many individuals associated with your centre performed cordocentesis? (Please do not include Obstetric residents in these figures.):
	Obstetricians
	Non-Obstetrician physicians
В.	CLIENT BASE
18.	In 1990, what was your centre's catchment/service area and population?
19.	In 1990, what proportion of your clients were from rural/distant locales and what proportion were urban/local? (This need only be answered if patient postal codes were not available for analysis or the data were largely incomplete.)
20.	Are there any genetic disorders with an unusually high incidence in your catchment area ie: haemoglobinopathies?
21.	Are there any large clusters of ethnic groups in your area that are at a higher risk than the general population for a particular genetic disorder(s)?
22.	Have you noticed a significant <u>increase in demand</u> for prenatal diagnosis in the past five years?

J	Have there been any major <u>shifts in referral patterns</u> in the pas years?
_	If so, what?
	Are there expected to be any major increases in the numb women being referred in the next few years? Yes/No (Y/N
	How have the trends in the above three questions been detect assessed?
-	
-	
-	
_	ACCESSIBILITY OF SERVICES
]	In 1990, at what age (at the time of delivery) was amniocen
]	ACCESSIBILITY OF SERVICES In 1990, at what age (at the time of delivery) was amniocen offered to women for advanced maternal age?: year In 1990, at what age (at the time of delivery) was CVS offer women for advanced maternal age?: years.
]	In 1990, at what age (at the time of delivery) was amniocer offered to women for advanced maternal age?: year In 1990, at what age (at the time of delivery) was CVS offer

	Please elaborate:
	1990, was prenatal counselling performed by Obstetricians and sindependent of the Genetics service? Yes/No (Y/N) [
	nat proportion of prenatal patients who proceeded to prenata ting were counselled by GPs or Obstetricians?
yoı	he prenatal counselling was done by an Obstetrician or GP, did in centre obtain copies of the patient information ie: pedigree? Yes/No (Y/N) [Comment:
 In	1990, did you offer genetic counselling/fetal assessment to al
pat ·	cients with high AFP?
In I	1990, was amniocentesis routinely offered to all women with high P?

	In 1990, did you offer genetic counselling/fetal assessment to all patients with an elevated risk for Down syndrome due to low AFP? Yes/No (Y/N) []
	Comment:
	In 1990, was amniocentesis routinely offered to all women with an age adjusted risk higher than that cutoff? Yes/No (Y/N) [] Comment:
	SERVICES OFFERED
111	ISPITITO:
	Were all referral groups treated in the same way ie: advanced maternal age versus single gene defect/neural tube defect? For
	Were all referral groups treated in the same way ie: advanced maternal age versus single gene defect/neural tube defect? For instance, was maternal age counselling left to GPs and Obstetricians
	Were all referral groups treated in the same way ie: advanced maternal age versus single gene defect/neural tube defect? For instance, was maternal age counselling left to GPs and Obstetricians
	Were all referral groups treated in the same way ie: advanced maternal age versus single gene defect/neural tube defect? For instance, was maternal age counselling left to GPs and Obstetricians while more complex situations were dealt with by Geneticists?
	Were all referral groups treated in the same way ie: advanced maternal age versus single gene defect/neural tube defect? Fo instance, was maternal age counselling left to GPs and Obstetricians while more complex situations were dealt with by Geneticists? How many prenatal clinics did you have each week?

In 1990, what information was requested at counselling ses your centre?	ssion
a) a pedigree of generations	
b) information re: general health of couple	
c) information re: any risk factors in this pregnancy d) any genetic disorders in the pedigree	
e) ethnic/racial background	
f) socio-economic status	
g) depended on the reason for referral	
Please elaborate:	
How were women scheduled for prenatal counselling?	
a) as soon as possible	
b) between and weeks' gestation c) whenever possible	
d) depended on the indication for prenatal diagnosis	
Please elaborate:	
Is the scheduling of the counselling session/procedure alterwomen/couples have to come in from a considerable distar	ed w
Yes/No (Y	
If yes, how?:	

What risk figures were quoted at the time of	counse	elling?	
Risk of a live-born child with Down syndr Risk of a live-born child with any chromo Risk of Down syndrome at the time of the Risk of any chromosome abnormality at t of procedure	some ab		ity [[
Other (Please specify)			
What was considered to be the optimal counselling and the actual procedure?			
	Numbe	er of day	<u>'S</u> :
amniocentesis			
CVS			
Were spouses/significant others encouraged	d to com	ne to:	
	Yes		No
counselling sessions	[]		[]
ultrasound			
			LI
amniocentesis CVS			
	[] nslation	ı in your	centr
CVS			

women determined to have an abnormal fetus?
a) after diagnosis, but prior to any decision as to whether or not to terminate:
b) after the decision to continue with or terminate the pregnancy:
c) after the birth of an affected child:
d) after a termination of pregnancy:
If the fetus was determined to be affected and the pregnancy was to be continued:
a) what counselling/literature/support groups were made available to the couple?
b) was the Genetics Centre involved in the medical management
of the pregnancy? Yes/No (Y/N) []
In 1990, were women required to agree to a termination of pregnancy as part of the condition for having amniocentesis/CVS if an abnormality was detected? Yes/No (Y/N) []

o d	n 1990, how many individuals in your catchment area would carry ut second trimester terminations of pregnancy following prenata iagnosis of a fetal abnormality? (Please do not include Obstetricesidents in these figures.):
	Obstetricians
	Non-Obstetrician physicians
t	n 1990, how were you set up to deal with first trimester erminations ie: were they widely available (number of hospitals able provide this service, performed on an in/out-patient basis?):
	n 1990, until what gestation were second trimester terminations for enetic reasons offered? weeks.
V	How were you set up to deal with second trimester terminations in vere they widely available, type of procedure, in/out-patient procedures, what ward was the woman put on?
	n 1990, did you maintain information about support groups regional/national/international) for different disorders?
V	Was this updated on a regular basis? Yes/No (Y/N) [
ŀ	How often do you review/update handout material?

67.	Do you update health professionals about changes in services offered or newly developed prenatal diagnostic techniques?
68.	In 1990, did you bank DNA from families with biochemical/molecular disorders for future analysis/interpretation? Comment:
MS-A	AFP Determination:
69.	Is MS-AFP determination offered as a population screening test in your centre/province? Yes/No (Y/N) []
70.	Indications for MS-AFP determination: Offered routinely during pregnancy [] History of maternal diabetes [] Family history of neural tube defects [] Other (Please elaborate on next page) []
7 1.	In 1990, between what gestations was MS-AFP testing recommended? Lower limit weeks, upper limit
72.	What control values were used? In-house normative data [] Reference laboratory values [] Kit data [] Other [] Please elaborate:
73.	What did you consider a "high" MS-AFP value?: greater than multiples of the median (MOM)
74.	What did you consider a "low" MS-AFP value?: less thanMOM

In the	1990, what written information was distributed to women about AFP test at their doctor's office (ie: prior to having the test)?
In 1	1990, who counselled women with abnormal MS-AFP values?
	s MS-AFP counselling performed in clinics or according to a errent schedule?
How	w soon after counselling for high/low MS-AFP values was niocentesis usually performed ie: ASAP, within a week?
tes	1990, did you offer additional maternal serum screening ie: triple ting - AFP, estriol, beta HCG? Yes/No (Y/N) [f so, under what circumstances?
	w did you determine which samples of amniotic fluid required tylcholinesterase determination?
In ser	1990, how did you follow up women with an "elevated" materna

ocentesis also have MS Yes/No (Y/N atesis for high MS-AFP Yes/No (Y/N atesis for a family history as determined? Yes/No (Y/N
tesis for high MS-AFP Yes/No (Y/N ntesis for a family histors des determined?
ntesis for a family histors determined?
s determined?
Yes/No (Y/N
ere you using (eg: <i>in</i> ?
e you using (eg: direct,

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96.	What, if any, special provisions are available for women requesting prenatal diagnosis who were found to have a twin pregnancy?
97.	In 1990, what protocol was in place for requesting a termination of pregnancy if an affected fetus was found?
Ultra	sound/Amniocentesis/CVS:
98.	Were all diagnostic ultrasounds performed in a fetal assessment unit that was associated with the Genetics centre?
99.	If no, was there any follow-up of ultrasounds that had been requested and were performed independently?
100.	In 1990, if women opted for ultrasound alone what gestation was this scheduled for?
	weeks (lower limit), (upper limit).
101.	In 1990, what was the maximum number of procedures your centre was able to accommodate per week?
	amniocentesis CVS
102.	In 1990, at what gestation was amniocentesis usually scheduled for?
	weeks (lower limit)
	weeks (upper limit)

103.	What factors limited the number of amniocenteses performed each week?
	lack of demand
	unable to schedule counselling
	unable to schedule procedure []
	laboratory restrictions:
	lack of microscopes
	lack of incubator space
	delays in film development [] lack of technicians []
	other []
	Please elaborate:
	Please elaborate.
104.	How did you deal with a potential oversubscription to this service?
	Restrict services and refer patients to other centres []
	Prearranged, reciprocal arrangements with other centres []
	Perform the procedure but ship the fluid elsewhere
	Other
	Please elaborate:
105.	In 1990, how did you deal with late referrals to Genetics ie: women referred after 24 weeks' gestation with advanced maternal age as the indication for referral?
106.	In 1990, at what gestation was CVS usually scheduled for?
	weeks (lower limit)
	weeks (upper limit)
107.	Was a 16-18 week follow-up ultrasound post-CVS standard practice in your centre?

108.	What factors limited the number of CVS procedures performed each week?
	lack of demand [] unable to schedule counselling [] unable to schedule procedure []
	laboratory restrictions:
	lack of microscopes [] lack of incubator space [] delays in film development [] lack of technicians []
	other []
	Please elaborate:
109.	How did you deal with a potential oversubscription to this service? Restrict services and refer patients to other centres []
	Prearranged, reciprocal arrangements with other centres Perform the procedure but ship the fluid elsewhere Other []
	Please elaborate:
110.	How was CVS offered at your centre (ie: first come, first served), what options were given to women who requested the procedure when there were no longer any slots available?
111.	Did physicians performing amniocentesis/CVS outside the centre have special arrangements with the cytogenetics laboratory?
	If yes, what:

In 1990, how did you deal with samples that arrived with little or warning?
EOLI OW UP
FOLLOW-UP
In 1990, did you review/audit your prenatal services? Yes/No (Y/N)
How often was this done?
What type of information was reviewed? (Please attach a lis possible)
Was there an institutional mechanism for review ie: a Prena Diagnosis Committee? Yes/No (Y/N)
What mechanisms were in place for the follow-up of patients what had prenatal diagnosis?
had had prenatal diagnosis?
What steps were taken to follow up:
had had prenatal diagnosis?

	b) Spontaneous abortions	following prena	tal diagnosis:	
	c) Therapeutic abortions:			
18.	What mechanisms were in p had MS-AFP screening (were oup)?			
19.	In 1990, did your province have the so, what interaction did		Yes/No (Y/N) []
uali 20.	ity Control Mechanisms: For what aspects of your property control mechanisms?	orenatal service	did you have	e quality
	Cytogenetics AFP Biochemical Molecular		Yes [] Yes [] Yes [] Yes []	No [] No [] No []
21.	Were there written guidelines	s for each of the	ese?	
	Cytogenetics AFP Biochemical Molecular		Yes [] Yes [] Yes [] Yes []	No [] No [] No []
	If no, please elaborate:			

Who determines this? Was there any lay/patient/consurrepresentation in this process?
In general, what was the decision-making process in each centand how were local policies established?
How were individual decisions made with respect to particular cathat may be at variance with centre policy ie: physic
judgment/referral to a committee/centre director?
LEGAL/ETHICAL CONCERNS
LEGAL/ETHICAL CONCERNS Are Genetic Counsellors and Ph.D.s who counsel insured?

Comment:

Outreach Questions

	TRE CODE:
Did you have an Outreach program in 1990?	Yes/No (Y/N) []
What area was served by this program?	
How often were the satellite clinics visited?	
Did you make a separate trip to each? If they were combined how was this done?	. Yes/No (Y/N) []
How long would each trip be scheduled for (numnumber of clinics)?	iber of days and total
	e participated in the

IC 1:	d /h arr did thar li	aiaa with the Co	metics of	inio?
ir yes, di	d/how did they li 	aise with the Go		mic:
How many	patients would b	e seen in each o	linic?	
Of these, w	hat proportion w	ere coming for p	orenatal o	counselling
Who couns	selled the <u>routing</u> s, local Obstetricia	e prenatal pations?	ents: Ou	itreach ni
** 1	ild a mamatal con	44.		
Were pedig	rees, family histor		les obtair	ned prior to
Were pedig session? . Commer	rees, family histor	ries, blood samp	les obtair	ned prior to
Were pedig session? . Commer	rees, family histor	ries, blood samp	les obtair	ned prior to
Were pedig session? . Commer What type facilities?	rees, family histor nt: e of prenatal tes	ries, blood samp	les obtair Yes	the Outr
Were pedig session? . Commer	rees, family histor nt: e of prenatal tes	ries, blood samp	les obtair Yes able at Yes	the Outr

_	
	as there a protocol in place for shipping samples ie: how wa ain centre notified?
	oproximately how many women were seen for probunselling/diagnosis through Outreach in 1990?
	ow many samples were generated by the Outreach progra
H	ow was the reporting of results handled?
H	ow was patient follow-up organized?
w	ho provided follow-up counselling, if necessary or requested
	1990, how many individuals at <u>each</u> of the regional outposts volved in Genetics liaison?

propo	the Outreach positions full time in 1990? If not, rtion of their time was spent doing this work (please panswer in terms of "Full-Time Equivalents")?
	was the Outreach program funded, was it funded sepa the "core" Genetics program?
progr	990, was there an advisory committee for the Out
	re was an advisory committee, how often did it meet?
	there representatives from disciplines other than Genet ommittee?

	nere was no committee, how were changes to the operation ocol made?
How	were physicians practising in your Outreach area kept inform
	ny new developments ie: newsletter, educational program?

Appendix 2. Mail-In Questionnaire

ASSESSMENT OF PRENATAL SERVICES OFFERED IN A GENETIC SERVICE SETTING

Please note: Those questions that require additional space should be completed on the attached sheets provided. Please photocopy the sheets if more are required.

A. CLIENT BASE

We would like the following information from each patient counselled about prenatal diagnosis in your Genetics centre in 1990 downloaded from the computer: postal code and, if possible, date of birth, reason for referral and referring physician. If postal code information is not available, then the closest city/town, township name or health region would be a useful alternative. If you enter a standard set of reasons for referral, please could you enclose a copy of this list. These data can be presented in any of the

following formats: ASCII, dBASE, PCFile, Lotus 1-2-3 and on any current MS-DOS media - 3.5", 5.25", dual-density or high-density disks. If you archive the data file, please include a copy of the archiving program.

General Practitioners Obstetricians Other physicians Community clinics Women's health services Maternal serum alpha-fetoprotein (MS-AFP) Fetal assessment unit(s) Self-referred Other - public health nurses, outreach, etc. Total: Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years over 44 years Total: * Age at (expected) time of delivery (Many centres will not be able to respond to questions 3-5	1.	Sources of 1990 prenatal patient referrals:	Number of	patients:
Other physicians Community clinics Women's health services Maternal serum alpha-fetoprotein (MS-AFP) Fetal assessment unit(s) Self-referred Other - public health nurses, outreach, etc. Total: Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years Total: * Age at (expected) time of delivery		General Practitioners		
Community clinics Women's health services Maternal serum alpha-fetoprotein (MS-AFP) Fetal assessment unit(s) Self-referred Other - public health nurses, outreach, etc. Total: Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years Total: * Age at (expected) time of delivery		Obstetricians		
Women's health services Maternal serum alpha-fetoprotein (MS-AFP) Fetal assessment unit(s) Self-referred Other - public health nurses, outreach, etc. Total: Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years Total: * Age at (expected) time of delivery		Other physicians		
Maternal serum alpha-fetoprotein (MS-AFP) Fetal assessment unit(s) Self-referred Other - public health nurses, outreach, etc. Total: Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 20-24 years 30-34 years 35-39 years 40-44 years over 44 years over 44 years Total: * Age at (expected) time of delivery		Community clinics		
Fetal assessment unit(s) Self-referred Other - public health nurses, outreach, etc. Total: Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 20-24 years 30-34 years 35-39 years 40-44 years over 44 years Total: * Age at (expected) time of delivery		Women's health services		
Self-referred Other - public health nurses, outreach, etc. Total: Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years * Age at (expected) time of delivery Total: * Age at (expected) time of delivery		Maternal serum alpha-fetoprotein (MS-AFP)		
Other - public health nurses, outreach, etc. Total: Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years * Age at (expected) time of delivery Total: * Age at (expected) time of delivery		Fetal assessment unit(s)		
Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years * Age at (expected) time of delivery		Self-referred		
Number of individuals multiply referred 2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years * Age at (expected) time of delivery		Other - public health nurses, outreach, etc.		
2. Age distribution of women referred for prenatal counselling in (If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years rotal: * Age at (expected) time of delivery			Total:	
(If possible, please attach frequency data for those women or years using yearly intervals ie: the number of women refer 1990 who will be 35 years of age at the time of delivery.) Woman's age* under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years * Age at (expected) time of delivery		Number of individuals multiply referred		
under 15 years 15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years * Age at (expected) time of delivery	2.	(If possible, please attach frequency data for years using yearly intervals ie: the number	those wome of women re	n over 35 eferred in
15-19 years 20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years Total: * Age at (expected) time of delivery		Woman's age*	Number	of women
20-24 years 25-29 years 30-34 years 35-39 years 40-44 years over 44 years Total: * Age at (expected) time of delivery		under 15 years		
25-29 years — 30-34 years — 35-39 years — 40-44 years — over 44 years — Total: * Age at (expected) time of delivery		15-19 years		
30-34 years		20-24 years		
35-39 years — 40-44 years — over 44 years — Total: * Age at (expected) time of delivery		25-29 years		
40-44 years — over 44 years — Total: * Age at (expected) time of delivery		30-34 years		
over 44 years Total: * Age at (expected) time of delivery		35-39 years		
* Age at (expected) time of delivery		40-44 years		
* Age at (expected) time of delivery		over 44 years		
 * Age at (expected) time of delivery (Many centres will not be able to respond to questions 3-5 			Total:	
		 * Age at (expected) time of delivery (Many centres will not be able to respond to 	questions	3-5)

3.	Level of education of women referred for prenatal counselling in 1990: Number of women
	Completed high school
	University graduate
	Post-graduate training
	Total:
4.	Gravid status of women referred for prenatal counselling in 1990:
	Number of women
	First pregnancy ———
	Second pregnancy
	Third pregnancy ————
	Fourth pregnancy or more
	Total:
5.	Parity of women referred for prenatal counselling in 1990:
	Number of women
	Childless ———
	One child ———
	Two children ———
	Three or more children ————
	Total:
B.	ACCESSIBILITY OF SERVICES
1.	Are requests for invasive prenatal diagnostic procedures ever refused?
	If yes, what do you consider valid reasons for refusing such requests? Please use attached sheet.

2.	In 1990, how many patients requesting invasive prenatal diagnostic procedures were refused testing?		
	Please document the reasons the procedures were denied and the number in each group. Please use attached sheet.		
3.	In 1990, how many requests for invasive prenatal diagnostic procedures were solely motivated by anxiety?		
	How many of those were refused testing?		
	How did those that were <u>not</u> refused differ from the above? Please use attached sheet.		
4.	In 1990, how many requests for invasive prenatal diagnosis solely for non-medical reasons were obtained ie: sex selection?		
	How many were refused?		
	How did those that were <u>not</u> refused differ from the above? Please use attached sheet.		
5.	In 1990, if a request for an invasive procedure was, for whatever reason, refused was an alternative offered? ie: a different procedure, referral to another centre		
	Comment:		
c.	SERVICE STATISTICS		
Pren	atal Referrals:		
1.	Number of prenatal referrals for each class of indication in 1990: Where there were cases with multiple indications, please record the indication with the highest risk and note that more than one risk factor was present ("Number referred for more than one reason").		
	Number of referrals		
	Advanced maternal age		
	Previous chromosome abnormality		

	Parental chromosome abnormality
	Relative (other than parent or offspring) with Down syndrome or other chromosomal abnormality
	High risk of chromosome abnormalities/neural tube defect or other anomalies based on serum AFP levels alone, or combined with other biochemical markers
	Previous neural tube defect
	Inborn error of metabolism
	Other single gene disease
	Genetic disorder with identifiable chromosome marker or abnormality
	Maternal/paternal irradiation
	Abnormal ultrasound
	Teratogen exposure
	Sex for medical reasons (eg: X-linked diseases)
	Other (Please specify on attached sheet)
	Total:
	Number referred for more than one reason
2.	Average gestational age of women referred for genetic counselling: weeks. (Calculated by ultrasound [] or LMP [])
ou	nselling:
3.	Average gestational age of women receiving genetic counselling: weeks. (Calculated by ultrasound [] or LMP [])
ŀ.	Total number of prenatal diagnosis counselling sessions in 1990:
	Only the woman was present at the counselling session
	Both members of the couple present
	Unknown
	Sub-total:
	No-shows (ie: a Genetics chart was made up for the patient)
	Total:

5.	In 1990, the number of women counselled in 1990 by:				
	a) M.D. geneticistsb) Ph.D. geneticists				
	c) genetic counsellors				
	d) other individuals associated (R.N.s, etc.)	l with Gen	etics		
			Tota	al:	
6.	In 1990, number of women receiving amniocentesis/CVS withou undergoing formal counselling by Genetics in this pregnancy because they had been counselled in a previous pregnancy:			pregnancy	
7.	Number of women eligible for receive the procedure:	amniocen	tesis/CVS w	ho did not	
	Reasons for missed procedure:	CVS	<u>Amnio</u>	<u>Total</u>	
	Dead/disorganized fetus found at time of procedure				
	Twin pregnancy				
	Miscarried prior to the procedure				
	No-show/decided against the procedure				
	Other (please elaborate)				
Ultra	sound:				
8.	How many diagnostic ultrasound 1990?:	ds were re	equested by	Genetics in	
	Indication for ultrasound:				
	Ultrasound due to an MS-AFP result				
	Amniocentesis/CVS declined b	y patient			

	Increased risk of structural malformation
	Other (Please elaborate on attached sheet)
	Total:
IS-/	AFP Determination:
).	In 1990, how many pregnant women were screened using MS-AFP?
Ο.	In 1990, how many prenatal MS-AFP tests were done in your centre?
1.	How many women were ascertained with at least one abnormal AFP value?
2.	Number of counselling sessions for abnormal AFP values (if separate from the above):
3.	In 1990, how many women underwent amniocentesis as a result of a "high" MS-AFP result?
	How many of these women would have been over the age of 35 (at the time of delivery)?
4.	In 1990, how many women underwent amniocentesis as a result of a "low" MS-AFP result?
	How many of these women would have been over the age of 35 (at the time of delivery)?
5.	How many of the women from questions 13 and 14 had previously declined amniocentesis or CVS?
\mr	niocentesis:

Number of amniocenteses performed for each class of indication in 16. 1990:

If there were cases with multiple indications, record the indication with the highest risk and note that more than one risk factor was present ("Number referred for more than one reason").

17.

<u>Indication</u>	<u>I</u>	Number of amniocenteses
Advanced maternal a	age	
Previous chromosom	e abnormality	
Parental chromosom	e abnormality	
Relative (other than property of the Down syndrome or o		
High risk of chromos defect or other anom levels alone, or comb markers (see questio	alies based on servined with other bid	um AFP
Previous neural tube	defect	
Inborn error of metal	bolism	
Other single gene dis	sease	
Genetic disorder with or abnormality	n identifiable chron	nosome marker
Maternal/paternal ir	radiation	
Abnormal ultrasound	đ	
Teratogen exposure		
Sex for medical reason	ons (eg: X-linked d	liseases)
Ambiguous CVS resu	ılt/failed CVS	
Other (Please specify	on attached sheet	
		Total:
	(Total # o	of women having amniocentesis)
Number referred for	more than one reas	son
Number of repeat am	nniocenteses (see q	uestion 22)
Multiple gestations	Indication	Number of sacs tapped
Twin pair A		
Twin pair B		
(If more space is requ	ired, please attach	an extra sheet)
In 1990, number of worto anxiety:	men/couples reque	esting amniocentesis due

18. In 1990, at what gestation was amniocentesis usually per		
	Median: weeks Range: Lower limit to Upper limit weeks.	
	(Dates calculated by ultrasound [] or LMP [])	
19.	In 1990, how many amniocenteses were performed:	
	Prior to 13 weeks' gestation	
	Between 13 and 15-6/7 weeks' gestation	
	Between 16 and 19-6/7 weeks' gestation	
	Between 20 and 23-6/7 weeks' gestation	
	24 weeks' gestation or greater	
	(Dates calculated by ultrasound [] or LMP [])	
20.	What was the average "turnaround time" from the date of the procedure until the date the karyotype was made available? days	
21.	In 1990, how many amniocenteses were performed following CVS due to:	
	Failure to obtain an adequate sample with CVS	
	Failure to obtain a laboratory result with an adequate sample from CVS	
	An equivocal laboratory result from CVS	
	Confirmation of an abnormal laboratory result from CVS	
22.	Number of repeat amniocenteses performed and the reasons they were repeated (ie: unable to obtain fluid, culture did not grow, equivocal result). This should <u>not</u> include amniocenteses that were deferred because of inaccurate gestational ages. $n = $ Please use attached sheet.	
23.	Number of women for whom a repeat amniocentesis was appropriate who declined the test:	
24.	In 1990, how many amniocenteses were performed outside the centre and fluid submitted for analysis?	

What were the indications for these amniocenteses? Please use attached sheet.

25. In 1990, how many amniocenteses were performed within your centre and the fluid processed elsewhere due to insufficient laboratory resources at your centre? Please do not count those samples that were sent to another centre for analysis where the expertise was not available in your own:

01	10	
# - N	-	-
No. N		-

26.	In 1990, how many women were referred early enough for CVS counselling (ie: under 13 weeks' gestation)?
27.	Average gestational age of women referred early enough for CVS counselling: weeks
28.	Of those women referred early enough for CVS counselling and for whom it was an appropriate test, how many chose:
	CVS
	Amniocentesis
	Ultrasound only
	No testing
29.	How many CVS procedures were performed in 1990?
	Number of transcervical procedures:
	Number of transabdominal procedures:
	Total:

30. Number of CVS procedures performed for each class of indication in 1990:

If there were cases with multiple indications, record the indication with the highest risk and note that more than one risk factor was present.

	<u>Indication</u>	Number of procedures
	Advanced maternal age	
	Previous chromosome abnormality	
	Parental chromosome abnormality	
	Relative (other than parent or offspring) we Down syndrome or other chromosomal abnormality	rith
	Inborn error of metabolism	
	Other single gene disease	
	Genetic disorder with identifiable chromo or abnormality	some marker
	Maternal/paternal irradiation	
	Teratogen exposure	
	Sex for medical reasons (eg: X-linked dis	eases)
	Repeat CVS (see question 34)	
	Other (Please specify on attached sheet)	
		Total:
	Number referred for more than one reaso	n
31.	In 1990, number of women/couples request	ting CVS due to anxiety:
32.	In 1990, at what gestation was CVS usually	y performed?
	Median: weeks Range: Lower limit to Upper limit (Dates calculated by ultrasound [] or LN	weeks.
33.	What was the average "turnaround time" fr date karyotype was made available?	om date of procedure to days.
34.	Number of repeat CVS tests performed and repeated (ie: unable to obtain villi, culture	d the reasons they were did not grow).
	This should <u>not</u> include procedures that we inaccurate gestational ages. $n = $ Pl	vere deferred because of lease use attached sheet.
35.	Number of women for whom a repeat appropriate who declined the test:	CVS test was deemed

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follow-up ultrasound post-CVS:

36.

37.

	tissue submitted for analysis?:	
	What were the indications for these CVS procedures? Please use attached sheet.	
38.	In 1990, how many CVS tests were performed within your centre and the sample sent elsewhere due to insufficient laboratory resources at your centre? Please do not count samples that were sent out for technical expertise not present in your centre:	
Bioch	nemical Prenatal Testing:	
39.	In 1990, how many tests for prenatally diagnosable biochemical disorders were analyzed by the laboratory with which you are affiliated? (Please do not include MS-AFP tests):	
	Document the reason for the test and the number in each category. Please use attached sheet.	
40.	In 1990, for what prenatally diagnosable biochemical disorders was the laboratory with which you are affiliated able to offer prenatal testing? Please use attached sheet.	
41.	In <u>1991</u> , for what prenatally diagnosable biochemical disorders can the laboratory with which you are affiliated offer prenatal testing? Please use attached sheet.	
42.	In 1990, what proportion of your biochemical service was oriented toward:	
	prenatal diagnosis % (n =)	
	carrier screening % (n =)	

Number of dead or disorganized fetuses found at the 16 week

(Note: not all centres may perform follow-up ultrasound after CVS)

In 1990, how many CVS tests were performed outside the centre and

Molecular Prenatal Testing:

43. In 1990, how many molecular tests for prenatally diagnosable genetic disorders were analyzed by the laboratory with which you are affiliated?

Document the reason for the test and the number in each category. Please use attached sheet.

- 44. In 1990, for what disorders was the laboratory with which you are affiliated able to offer prenatal molecular testing? Please use attached sheet.
- 45. In <u>1991</u>, for what disorders can the laboratory with which you are affiliated offer prenatal molecular testing? Please use attached sheet.
- 46. In 1990, what proportion of your molecular service was oriented toward:

prenatal diagnosis ______ % (n = _____)
carrier screening _____ % (n = ____)

Other:

- 47. In 1990, how many prenatal tests other than amniocentesis, CVS, ultrasound, MS-AFP were performed ie: cordocentesis? Please document the number and types of tests performed, the indication for the test, procedure risks quoted and any complications that arose.
- 48. In 1990, if a couple opted to terminate a pregnancy due to an affected fetus, what was the average length of time that lapsed between the diagnosis of an affected fetus and the termination of pregnancy: _____ days.

D. OUTCOMES

This section deals with complications of pregnancy and abnormal outcomes after prenatal diagnosis. Please complete the attached table: "1990 abnormal pregnancy outcomes and complications of pregnancy following prenatal diagnosis."

1.	In 1990, did you have any false positive results from prenatal diagnosis ie: a genetic abnormality was diagnosed prenatally but the fetus/live born was subsequently found to be normal?		
	If yes, please complete the attached sheet.		
2.	In 1990, did you have any false negative results from prenatal diagnosis ie: abnormalities that should have been diagnosed prenatally were missed? This would also include cases where the sex of the infant was determined incorrectly Yes/No (Y/N) []		
	If yes, please complete the attached sheet.		
E.	STAFFING		
perso	responding to the questions in this section involving number of nnel, please could you phrase your response in terms of "Full-Time ralents."		
1.	In 1990, how many M.D.s were involved in prenatal counselling?		
2.	How many were accredited by the CCMG?		
	In what specialities? <u>Number of individuals</u> :		
	Clinical Genetics		
	Cytogenetics		
	Molecular Genetics		
	Biochemical Genetics		
	How many were accredited by the RCPS and in what specialities?		

In what capacity? Prenatal counselling Cytogenetics Molecular diagnosis Biochemical diagnosis	Number of individual
MS-AFP determination	
Other (Please specify)	
How many of the Ph.D.s were a	accredited by the CCMG?
In what specialities?	Number of individua
Medical Genetics	
Cytogenetics	
Molecular Genetics	
Biochemical Genetics	
In 1990, how many Genetic prenatal service?	Counsellors were involved in t
Of these, how many had:	
Formal training in a Genetic	Counselling program?
A master's degree in genetic rather than counselling focu	
Nursing degree?	
R.N. diploma?	
Other (Please specify below)	

Genetic Counsellor

Nurse/R.N.

c) Testing:		
Cytogenetics Biochemical/Molecular		Technologist
d) Miscellaneous:	FII.D	
·		
Outreach Support staff		
What level of funding are ye	ou using for this	s estimate?
Optimal/ideal funding Current levels		[]
If they are assessed at curre	ent levels then h	now are these assessed?:
Ideal Adequate Inadequate		
Comment:		
BUDGET		
In 1990, was the budget fo general genetics service bu		
In 1990, how was the budg	get for the prena	tal service arrived at?

G.

1.

2.

What was the cost of the MS-AFP program?	
Was this included in the prenatal budget or was it fu separately?	nded
Does any part of the <u>service</u> funding for prenatal diagnosis of from research grants?	
Was the prenatal genetics service globally funded from a Geneservice budget or was the funding allotted on a fee per patient b	
In your centre how were priorities set?	
Within the genetics service (ie: prenatal diagnosis versus other services)	[]
Within your institution/hospital (ie: between the needs of the genetics service and other demands)	[]
Funding from the hospital global budget	[]
Funding from the Ministry of Health to a Provincial Genetic Program	[]
Other (Please comment below)	[]
At what level does your centre make its case for more funding	?
Government [] Institution [] Department [] Other []	
Please comment:	

Procedure Costs

1990 costs:

Cost of genetic counselling

We need at least laboratory costs, and the balance prorated between professional fees and genetic counselling for other reasons. Where possible this should be apportioned to specific areas.

Laboratory costs	
Procedure costs	
- amniocentesis	
- CVS	
- diagnostic ultrasound	
Shipping costs for samples sent elsewhere for interpretation	
Personnel Costs	
How were M.D.s involved in the service remunera	ated?
salaries sessional payments from global budget fee for service	[] [] []
Was the method of remuneration considered ap service provided?	
Comment:	Yes/No (Y/N) []
How were Ph.D.s involved in the service remuner	rated?
salaries sessional payments from global budget fee for service	[]

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Was the method of remuneration considered appropriate for the type of service provided? \dots Yes/No (Y/N) []
Comment:
How were genetic counsellors remunerated?
How were other professional staff remunerated?
If clients did not have health insurance, how was payment for services arranged?
If a procedure was not available in your area how were patients referred and payment arranged?

Appendix 3. Genetics Centres in Canada Offering Prenatal Diagnosis

BRITISH COLUMBIA

Vancouver

CCMG Accreditation: Yes: 1981, 1987

Director: Dr. J. Friedmann, M.D., Ph.D., FCCMG, FRCPC

Maternal Serum AFP Studies: Yes; all chorionic villus sampling and genetics prenatal patients have maternal serum AFP tests. Maternal serum AFP is also offered for maternal anxiety (along with an ultrasound) and teratogen exposure. In the community, some G.P.s and Obstetricians also screen their patients.

Computerized Data Base: Partial; Customized dBASE IV application

Client Base: Population of British Columbia; population 3.1 million

Region Served: Province of British Columbia

Associated Laboratories:

Cytogenetics: Yes; three laboratories in Vancouver (Children's Hospital, Vancouver General, Royal Columbian Hospital). All directors FCCMG; all laboratories report through Department of Pathology.

Molecular: Yes; part of Genetics, reporting through Department of Pathology.

DNA Banking: Yes

Biochemical: Yes; reporting through Children's Hospital, Director FCCMG.

Diagnostic Ultrasound: Majority at Grace Hospital. Some private laboratories and other hospital units.

Outreach: Yes; serving Thompson-Okanagan district with clinics in Kamloops, Kelowna, Vernon, Victoria, and Quesnel. Frequency bi-monthly, Quesnel annually.

Comments: A large and complex centre, organized through the Department of Medical Genetics at the University of British Columbia. The laboratories performing the testing required by the prenatal diagnosis program are located in various hospitals, and all report through the Department of Pathology. Much of the advanced maternal age counselling is done by obstetricians and G.P.s in the community and the clients never see a geneticist. Most procedures (amniocentesis, chorionic villus sampling, and cordocentesis) are performed at the Grace Hospital. Specific obstetricians associated with the genetics centre were designated to perform the procedures (amniocentesis — 7; chorionic villus sampling — 2; cordocentesis — 4).

Victoria

CCMG Accreditation: No

Director: None

Maternal Serum AFP Studies: None locally; all samples sent to Vancouver. Used primarily for clients at high risk for neural tube defects.

Computerized Data Base: No Client Base: 0.5 million population Region Served: Vancouver Island

Associated Laboratories:

Cytogenetics: Yes; not accredited, no professional director. In 1990, sign-off for cytogenetic analysis was done by Vancouver/Calgary.

Molecular: No

DNA Banking: No Biochemical: No

Diagnostic Ultrasound: Medical imaging, Victoria General

Hospital.

Outreach: No

Comments: This is a small centre, which lost both its medical geneticist and cytogeneticist in 1990. Support was provided by the medical genetics centre in Vancouver for genetic counselling, and cytogenetic analyses by laboratories in Calgary and Vancouver. Only amniocentesis was performed, primarily in the antenatal unit of the Victoria General Hospital. Most advanced maternal age counselling was performed by an obstetrician or family physician. An outreach clinic from Vancouver is now held every two weeks to see complex cases.

ALBERTA

Calgary

CCMG Accreditation: Yes; 1981, 1987

Director: Dr. Brian Lowry, M.D., D.Sc., FRCPC, FCCMG

Maternal Serum AFP Studies: Yes; done in Biochemistry and reporting through the Department of Pathology. Maternal serum AFP offered for a family history of neural tube defects, teratogen exposure (e.g., Valproic acid), maternal anxiety (as a Down syndrome screen), and upon patient request.

Computerized Data Base: Yes; Rbase 5000

Client Base: Population 1.2 million

Region Served: Alberta south of and including Red Deer;

southeast British Columbia

Associated Laboratories:

Cytogenetics: Yes; reports to director of genetics centre; laboratory director FCCMG. Funded by the Alberta Hereditary Diseases Program; amniocentesis and chorionic villus sampling funded by Laboratory Medicine.

Molecular: Yes; reports to director of genetics centre. Not affiliated with Laboratory Medicine.

DNA Banking: Yes

Biochemical: Limited; most samples submitted elsewhere.

Diagnostic Ultrasound: Available at Peter Lougheed Centre and Foothills Hospital. No formal relationship between Genetics and the Obstetric departments at these hospitals. Coordination is through the prenatal diagnosis coordinator.

Outreach: Extensive coverage: southern Alberta and eastern British Columbia. Mountainview, Lethbridge, Medicine Hat, Red Deer, and Drumheller visited monthly (primarily non-prenatal cases seen by geneticists). Urgent prenatal patients booked into Calgary. Advanced maternal age patients counselled by outreach nurse. Invasive testing available only in Calgary.

Comments: This centre is part of the Department of Paediatrics. The Cytogenetics Laboratory, although formally part of Laboratory Medicine, reports through the centre. The molecular laboratory is an integral part of the centre. In Calgary, chorionic villus sampling is done only for high-risk patients and is not available for advanced maternal age women. The centre has been performing early amniocenteses, outside the Canadian guidelines. as part of a pilot study. Approximately 1 000 such amniocenteses have been performed, although accurate data were not provided. Most counselling for advanced maternal age is now done in group sessions, and many advanced maternal age patients are not seen by Genetics but are counselled in the community by their physician. Procedures for patients referred to Genetics are carried out by obstetricians at either the Peter Lougheed Centre at Calgary General Hospital or Foothills Hospital. A significant amount of both counselling and procedures are undertaken in the community.

Edmonton

CCMG Accreditation: Yes; 1985, 1991 (provisional 1 year) **Director:** Dr. P. Ferreira, M.B.B.S., FRCPC, FCCMG

Maternal Serum AFP Studies: Limited; contracted to Winnipeg. Offered for a history of maternal diabetes, family history of neural tube defects, post-chorionic villus sampling, maternal anxiety, and possible teratogens (e.g., Valproic acid).

Computerized Data Base: Yes; SPIRES mainframe custom application

Region Served: Alberta north of Red Deer; northern British Columbia, the Yukon; and part of the Northwest Territories

Associated Laboratories:

Cytogenetics: Yes; reports to Chief of Laboratory Medicine; Director FCCMG; uses same data base.

Molecular: No
DNA Banking: Yes
Biochemical: No

Diagnostic Ultrasound: Private laboratory does most pre- and post-chorionic villus sampling scans. Fetal anomalies referred to Genetics.

Outreach: Extensive, through Alberta Hereditary Diseases Program, which covers all of Alberta north of Red Deer. The Peace River Health Unit, South Peace Health Unit, Fort McMurray and District Health Unit, and Alberta West Central Health Unit are visited bi-annually. Counselling of prenatal patients by outreach nurse. Maternal serum AFP and amniocentesis available at outreach sites.

Comments: This centre is part of the Department of Paediatrics at the University of Alberta. The CCMG has just reduced its accreditation from full to provisional for service delivery, primarily because of lack of communication between the Genetics Centre and the Cytogenetics Laboratory. Specific obstetricians are designated to perform prenatal testing (amniocentesis — 7; chorionic villus sampling — 2; cordocentesis — 4). Outreach program is extensive and well organized.

SASKATCHEWAN

Saskatoon

CCMG Accreditation: No

Director: Dr. M. Shokier, M.B., B.Ch., M.S., Ph.D., FCCMG,

FRCPC

Maternal Serum AFP Studies: No

Computerized Data Base: Yes; dBASE IV

Region Served: Central and northern Saskatchewan

Associated Laboratories:

Cytogenetics: Yes; part of Genetics Centre.

Molecular: No DNA Banking: No

Biochemical: Limited; most samples referred elsewhere.

Diagnostic Ultrasound: Available through the Department of Radiology at University Hospital. One private radiology clinic performs dating ultrasound.

Outreach: Yes; Regina visited monthly.

Comments: This is a relatively small centre which is not accredited by the CCMG. The division of Medical Genetics is part of the Department of Paediatrics. Clients seen by Genetics are usually those with more complex problems. Advanced maternal age counselling is largely done by physicians in the community.

There is a geneticist in private practice, and a perinatologist at Royal University Hospital. No routine maternal serum AFP screening is carried out in the province. Chorionic villus sampling is not available to women in Saskatchewan. There are no designated obstetricians who perform amniocenteses.

Regina

CCMG Accreditation: No

Director: None

Maternal Serum AFP Studies: Yes; done at Pasquaw Hospital. There is no maternal serum AFP screening program in Regina, but the Pasquaw Hospital processes both maternal serum and amniotic fluid AFP and the values are reported back to the patient's physician.

Computerized Data Base: No

Region Served: Southern Saskatchewan

Associated Laboratories:

Cytogenetics: Yes; no professionally accredited cytogeneticist.

Molecular: No DNA Banking: No Biochemical: No

Diagnostic Ultrasound: Not arranged by genetics.

Outreach: No

Comments: There is no genetics centre as such in Regina, only a cytogenetics laboratory. Complex cases are referred to Saskatoon. Amniocentesis is performed by community obstetricians who both counsel the client and perform the procedure.

MANITOBA

Winnipeg

CCMG Accreditation: Yes; 1985, 1990

Director: Dr. A.E. Chudley, M.D., FRCPC, FCCMG

Maternal Serum AFP Studies: Yes; provincial screening program **Computerized Data Base:** Yes; Paradox custom application

Client Base: 1.3 million population

Region Served: Manitoba, northwest Ontario

Associated Laboratories:

Cytogenetics: Yes; integral part of Genetics Centre, Director

FCCMG.

Molecular: No DNA Banking: Yes

Biochemical: Yes; limited. All prenatal samples sent out in 1990. Reporting through Clinical Chemistry and to the clinical

geneticist, and metabolic specialist.

Diagnostic Ultrasound: Yes; Fetal Assessment Unit and departments of Medical Imaging, Health Sciences Centre and St. Boniface Hospital.

Outreach: No

Comments: Clinical Genetics operates as a section of the Department of Paediatrics. Staff are all cross-appointed to the Department of Human Genetics. This centre provides a full range of tests, including amniocentesis, chorionic villus sampling (limit five per week), cordocentesis, detailed fetal assessment through ultrasound, and the only provincial maternal serum AFP screening program. No fluids are accepted from outside the centre. Amniocentesis and chorionic villus sampling are done only in the Fetal Assessment Unit.

ONTARIO

Hamilton

CCMG Accreditation: Originally accredited in 1982, and then in 1989. At present accredited in cytogenetics and biochemical genetics but not in clinical genetics.

Director: Dr. D. Whelan, M.D., FRCPC, FCCMG

Maternal Serum AFP Studies: Not part of centre, done in a separate hospital with no formal liaison with genetics centre. Maternal serum AFP would be offered to women who were having chorionic villus sampling, or who had had a previous neural tube defect. Pregnant diabetic patients would be screened by the high-risk obstetricians. Some physicians in the community routinely do maternal serum AFP screening on all pregnant patients.

Computerized Data Base: No

Region Served: Central and southwestern Ontario

Associated Laboratories:

Cytogenetics: Yes; but reporting to both Genetics and Laboratory Medicine.

Molecular: Yes; reporting to both Laboratory Medicine and Genetics. Haemoglobinopathies and phenylketonuria for province of Ontario.

DNA Banking: Yes

Biochemical: Yes; reporting to both Laboratory Medicine and Genetics. Amino acid and organic acid disorders for the province of Ontario.

Diagnostic Ultrasound: Regional referral centre; level II and level III ultrasounds, primarily at the Antenatal Diagnosis Unit.

Outreach: Yes; Sault Ste. Marie, visited three to four times per year. Prenatal patients usually not seen.

Comments: This centre does not at present have an accredited clinical geneticist. Full range of services except maternal serum AFP provided. Designated obstetricians (5 amniocentesis; 2 chorionic villus sampling) perform amniocentesis and chorionic villus sampling, although some fluids are accepted from elsewhere.

Kingston

CCMG Accreditation: Yes: 1990

Director: Dr. P. MacLeod, M.D., FCCMG, FRCPC

Maternal Serum AFP Studies: Yes; screening not available, maternal serum AFP offered primarily for a family history of neural tube defects. Director Ph.D. biochemist. Many centres send amniotic fluid samples here for acetylcholinesterase determination.

Computerized Data Base: Yes; dBASE II, also PC File

Region Served: Eastern Ontario, Peterborough, Frontenac, Hastings, Leeds, Grenville, Belleville, and Brockville

Associated Laboratories:

Cytogenetics: Yes; integral part of genetics centre, director CCMG. Reports also through Pathology.

Molecular: Yes; integral part of genetics centre, director FCCMG. Reports also through Pathology.

DNA Banking: Yes

Biochemical: Yes; director not CCMG-accredited. Integral part of genetics centre; reports also through Pathology.

Note: All laboratories are part of the Kingston General Hospital Laboratory system.

Diagnostic Ultrasound: Yes, at Kingston General Hospital. Abnormalities may or may not be referred to Genetics.

Outreach: Yes; Peterborough is a satellite of Kingston, Thunder Bay, Sudbury. Primarily not prenatal counselling, which is usually done locally by outreach nurses. Amniocentesis is available locally in Peterborough, Thunder Bay.

Comments: A well-integrated centre, with a full range of services, including amniocentesis, chorionic villus sampling (commenced Nov. 1990, one obstetrician), and diagnostic ultrasound. All amniocenteses and chorionic villus sampling tests are done at Kingston General Hospital. Laboratories integrated into Genetics although a part of Kingston General Hospital's Department of Laboratory Medicine.

London

CCMG Accreditation: Yes; 1981, 1988 Director: Dr. J. Jung, M.D., FCCMG, FRCPC

Maternal Serum AFP Studies: Not part of centre. Maternal serum AFP studies done through Biochemistry, laboratory director FRCPC. Maternal serum AFP is available at the discretion of the local physicians. It is also routinely available to any prenatal patients seen by the genetics centre.

Computerized Data Base: Yes; dBASE 2 application

Region Served: Southwestern Ontario

Associated Laboratories:

Cytogenetics: Yes; not part of centre, reports through Pathology, director FCCMG.

Molecular: No
DNA Banking: Yes

Biochemical: Yes; through the Children's Psychiatric Research

Institute.

Diagnostic Ultrasound: Level II and III ultrasounds done at

Children's Hospital of Western Ontario.

Outreach: Yes; Windsor (every two months), Sault Ste. Marie, and Thunder Bay (bi-annually). Few patients seen for prenatal diagnosis in outreach clinics. Maternal serum AFP and ultrasound available at outreach clinics, through referral to local hospitals. Results reported through Genetics and in cooperation with the obstetrician of record.

Comments: This centre serves southwestern Ontario delivering a full range of services, including genetic amniocentesis and chorionic villus sampling with designated physicians performing the tests. Independently of the genetics centre, about seven obstetricians in the area counselled patients and performed testing with minimal interaction with the centre.

Ottawa

CCMG Accreditation: Yes; 1983, 1989

Director: Dr. A. Hunter, M.D., C.M., FCCMG, FRCPC

Maternal Serum AFP Studies: Limited, not part of genetics centre. Performed in Immunology Laboratory at Ottawa Civic Hospital. High values reported to the genetics centre, then to physician. Low values not reported.

Computerized Data Base: Yes; dBASE III+ custom application

Client Base: 0.9 to 1.0 million population

Region Served: Ottawa, western Quebec, Sudbury, Smith Falls

Associated Laboratories:

Cytogenetics: Yes; integral part of genetics centre, director FCCMG.

Molecular: Yes; integral part of genetics centre, director FCCMG.

DNA Banking: Yes

Biochemical: No; samples referred elsewhere.

Diagnostic Ultrasound: Ottawa Civic and Ottawa General. High-risk and Obstetric ultrasound units. Close liaison with Genetics; combine rounds.

Outreach: Yes; North Bay (bi-annually), Sudbury (eight visits/year), and Timmins (quarterly). About 25 to 30 patients seen per visit by two geneticists. Prenatal patients not routinely seen on outreach visits; counselling usually done by outreach nurses. Maternal serum AFP and amniocentesis available only at Sudbury. Dating ultrasounds available but not level II. Non-routine patients referred to Ottawa, Toronto, North York. Communication of results and follow-up largely by outreach nurses.

Comments: A fully integrated centre performing a full range of services, except for biochemical diagnosis, which is referred elsewhere. Both genetic amniocentesis and chorionic villus sampling performed by designated obstetricians (6 genetic amniocenteses; 2 chorionic villus sampling).

Credit Valley

CCMG Accreditation: No

Director: Dr. S. Farrell, M.Sc, M.D., FCCMG, FRCPC

Maternal Serum AFP Studies: Yes; laboratory part of Laboratory Medicine. Most of the physicians in Credit Valley send maternal serum AFP samples, the physicians in the community may or may not.

Computerized Data Base: Yes; dBASE custom application

Client Base: 2.0 million population

Region Served: Peel County, Halton County, north to

Orangeville, and edge of Etobicoke

Associated Laboratories:

Cytogenetics: Yes; integral part of the genetics centre, director

FCCMG.

Molecular: No DNA Banking: No Biochemical: No

Diagnostic Ultrasound: Dating ultrasounds at Credit Valley. One private clinic also doing level II ultrasounds. Level III referred.

Outreach: Yes; Thunder Bay two to three visits per year. Maternal serum AFP, amniocentesis, and diagnostic ultrasound available.

Comments: This is a large general hospital and handles primarily advanced maternal age cytogenetic diagnoses. More complex noncytogenetic cases referred elsewhere.

North York

CCMG Accreditation: No

Director: Dr. P. Wyatt, Ph.D., M.D., FRCPC

Maternal Serum AFP Studies: Yes; laboratory an integral part of the genetics centre. All pregnant women seen at North York General Hospital are screened with maternal serum AFP.

Computerized Data Base: Yes; dBASE application; main file, prenatal and laboratory information; also subsidiary data bases

Client Base: 1.5 million population

Region Served: North of Highway 401, east to Ajax-Pickering, west to Highway 400

Associated Laboratories:

Cytogenetics: Yes; integral part of the genetics centre, director FCCMG.

Molecular: No DNA Banking: Yes Biochemical: No

Diagnostic Ultrasound: Most level II and level III ultrasounds done at North York; a few in private clinics.

Outreach: Yes; Sudbury, North Bay, Timmins, Orillia. Four visits per year to each centre by two geneticists. Routine prenatal patients counselled by outreach nurse. Amniocentesis and

maternal serum AFP available at prenatal centres. Fluids analyzed in either Sudbury or Ottawa.

Comments: This is a large general hospital serving the northern area of Metro Toronto. Its primary case load for prenatal diagnosis is advanced maternal age or other cytogenetic indications. Molecular and biochemical studies are referred to The Hospital for Sick Children.

Oshawa

CCMG Accreditation: No

Director: Dr. H.A. Gardner, M.D., FRCPC, FCCMG

Maternal Serum AFP Studies: Yes; laboratory part of Laboratory

Medicine

Computerized Data Base: Yes; Meditek **Client Base:** 0.7 to 0.75 million population

Region Served: Peterborough/Durham; Northumberland

Associated Laboratories:

Cytogenetics: Yes; integral part of genetics centre, director

FCCMG.

Molecular: No DNA Banking: Yes Biochemical: No

Diagnostic Ultrasound: Dating ultrasounds at Credit Valley. One private clinic also doing level II ultrasounds. Level III referred.

Outreach: No

Comments: This is a large general hospital serving Metro Toronto. Handles primarily advanced maternal age cytogenetic diagnoses. More complex non-cytogenetic cases referred elsewhere.

TORONTO PRENATAL DIAGNOSIS PROGRAM

Toronto General Hospital

CCMG Accreditation: Yes; 1981, 1988 **Director:** Dr. E. Hutton, Ph.D., FCCMG

Maternal Serum AFP Studies: Yes; no provincial screening program, but a significant proportion of births are screened.

Computerized Data Base: Yes; Carefile **Client Base:** 3 to 4 million population

Region Served: Amniocentesis patients come from south of the 401 highway. Molecular and other specialized cases are referred from all over Ontario.

Associated Laboratories:

Cytogenetics: Yes; report to Department of Pathology, director FCCMG accredited, laboratory is physically separate from the genetics centre.

Molecular: No — done at The Hospital for Sick Children.

DNA Banking: No — done at The Hospital for Sick Children.

Biochemical: No — done at The Hospital for Sick Children.

Diagnostic Ultrasound: Most are done at Toronto General Hospital; a few are done at the Fetal Assessment Unit. Numerous private ultrasound laboratories, and smaller ultrasound laboratories at other hospitals. All complex cases re-scanned at Toronto General Hospital.

Outreach: No

Comments: This hospital forms part of the University of Toronto Prenatal Diagnosis Program, doing most of the routine advanced maternal age counselling and cytogenetic diagnoses, and acting as a coordinating centre from which fluids are referred to other laboratories.

The Hospital for Sick Children

CCMG Accreditation: Yes; 1981, 1988

Director: Dr. R. Worton, Ph.D., FCCMG; Dr. J. Clarke, M.D.,

Ph.D., FCCMG, FRCPC

Maternal Serum AFP Studies: No, all maternal serum AFP done at Toronto General Hospital.

Computerized Data Base: No

Client Base: 3 to 4 million population

Region Served: All of Ontario, particularly Toronto. Mainly high risk for molecular and biochemical testing, or complex cytogenetic.

Associated Laboratories:

Cytogenetics: Yes; director FCCMG, an integral part of genetics centre.

Molecular: Yes; director FCCMG, integral part of genetics centre.

DNA Banking: Yes

Biochemical: Yes; director Ph.D. biochemist, integral part of genetics centre.

Diagnostic Ultrasound: All done at Toronto General Hospital.

Outreach: No

Comments: This hospital is part of the University of Toronto Prenatal Diagnosis Program. Most complex genetic cases are counselled at this hospital. All molecular, biochemical, and complex cytogenetic diagnoses are done in The Hospital for Sick Children laboratories.

Wellesley

CCMG Accreditation: No

Director: Dr. M. Shire, M.D., FRCPC

Maternal Serum AFP Studies: Yes; Department of Biochemistry, Wellesley Hospital. All women undergoing prenatal testing at the Wellesley will also have maternal serum AFP testing, although this

is not a formal province-wide screening program.

Computerized Data Base: Yes; Macwrite Client Base: 0.7 to 0.75 million population Region Served: Metro Toronto and Scarborough

Associated Laboratories:

Cytogenetics: Yes; director not FCCMG.

Molecular: No DNA Banking: No Biochemical: No

Diagnostic Ultrasound: Not done at this centre, referred to

Toronto General Hospital.

Outreach: No

Comments: This is a small centre performing genetic amniocentesis only, consists of an obstetric unit and a cytogenetic laboratory. Genetics support is available on an as-necessary basis. Two obstetricians perform genetic amniocentesis. Neither the centre nor its staff is accredited by the CCMG. Case load is primarily women referred for advanced maternal age. More complex cases referred elsewhere.

QUEBEC

Laval

CCMG Accreditation: No

Director: Dr. R. Gagné, M.D., FRCPC, FCCMG

Maternal Serum AFP Studies: Yes; no screening program, primarily for a family history of neural tube defects. Laboratory

part of Biochemical Genetics.

Computerized Data Base: Yes; partially

Region Served: Quebec City, Gaspé, north and east of Quebec City

Associated Laboratories:

Cytogenetics: Yes; integral part of Genetics, director FCCMG.

Molecular: Yes; integral part of Genetics. **DNA Banking:** Banks cells, not DNA.

Biochemical: Yes; integral part of Genetics.

Diagnostic Ultrasound: Usually done in local hospitals except for chorionic villus sampling ultrasound, all done at the Centre hospitalier de l'Université Laval.

Outreach: No

Comments: Amniocentesis done in community hospitals, chorionic villus sampling tests all performed at the Centre hospitalier de l'Université Laval by two obstetricians. Cordocentesis not performed at the Centre hospitalier de l'Université Laval.

McGill University

CCMG Accreditation: Yes; 1984, 1989

Director: Dr. M. Vekemans, M.D., FCCMG, FRCPC

Maternal Serum AFP Studies: No; processed in endocrinology, interpreted in Genetics. No screening program, high-risk cases only: history of maternal diabetes, family history of neural tube defects, post-chorionic villus sampling, and teratogen exposure.

Computerized Data Base: Yes; dBASE IV

Region Served: Montreal and suburbs; Val d'Or, northwest Quebec, Baffin Island, and Ungava Bay

Associated Laboratories:

Cytogenetics: Yes; integral part of Genetics although reporting through Pathology for administrative purposes, director FCCMG in 1990.

Molecular: Yes; director FCCMG.

DNA Banking: Yes

Biochemical: Yes; director FCCMG.

Diagnostic Ultrasound: Available at all McGill obstetric hospitals.

Outreach: No

Comments: Prenatal diagnosis program offers amniocentesis and chorionic villus sampling. Laboratories offer cytogenetic, molecular, and biochemical diagnoses. Twelve obstetricians

performed genetic amniocentesis, and one chorionic villus sampling. Four obstetricians performed cordocentesis.

University of Montreal

CCMG Accreditation: No

Director: Dr. L. Dallaire, M.D., Ph.D., FCCMG, FRCPC **Maternal Serum AFP Studies:** Yes; integral part of centre

Computerized Data Base: No

Region Served: Montreal/province of Quebec. One of three centres offering prenatal diagnosis in Quebec. Clients may be referred to most convenient.

Associated Laboratories:

Cytogenetics: Yes; director FCCMG. **Molecular:** Yes; director FCCMG.

DNA Banking: Yes

Biochemical: Yes; director FCCMG.

Diagnostic Ultrasound: Many private units. Most genetics

patients would have ultrasound at Ste. Justine.

Outreach: No

Comments: Hôpital Ste. Justine is a large, tertiary care teaching hospital. Genetics is part of the University Department of Paediatrics. Patients are referred from all over Quebec, although the primary case load comes from the Montreal francophone population. A full range of services is offered, including genetic amniocentesis, chorionic villus sampling, molecular, cytogenetic, and biochemical testing.

MARITIME PROVINCES

Halifax

CCMG Accreditation: No

Director: Dr. J.P. Welch, M.B., Ch.B., Ph.D., FCCMG, FRCPC **Maternal Serum AFP Studies:** Yes; no screening program; high risk and maternal anxiety. Part of laboratory services at Victoria Hospital. This laboratory does both maternal serum AFP and amniotic fluid AFP.

Computerized Data Base: Yes; dBASE III

Region Served: Referral centre for New Brunswick, Nova Scotia,

and Prince Edward Island

Associated Laboratories:

Cytogenetics: Yes; but not part of genetics centre; reporting through Laboratory Medicine, Izaak Walton Killam Hospital for Children. Director FCCMG.

Molecular: Yes; but part of Laboratory Services and Department of Pathology, Izaak Walton Killam Hospital for Children. Director not CCMG accredited.

DNA Banking: Yes

Biochemical: Yes; two laboratories — one is part of the Atlantic Research Centre (ARC) and the other is in the Izaak Walton Killam Hospital for Children. Reporting to ARC and Izaak Walton Killam Hospital for Children, respectively.

Diagnostic Ultrasound: Diagnostic imaging and through Obstetrics at the Grace Hospital.

Outreach: Yes; Charlottetown, Prince Edward Island; Saint John, Fredericton, and Moncton, New Brunswick. Bi-annual clinics. Most prenatal patients come to Halifax. Some amniocenteses done in Moncton and Saint John, none done in Prince Edward Island. All fluids sent to Halifax for analysis. Patients or physicians informed by telephone with option of attending a genetics clinic either outreach or Halifax.

Comments: A regional centre housed in the Atlantic Research Centre covering the whole of the Maritime provinces and partly funded from Prince Edward Island and New Brunswick. Provides a full range of services, including amniocentesis, chorionic villus sampling, and cordocentesis. Laboratories not part of centre; all report though Pathology or Laboratory Medicine of one or other teaching hospitals.

NEWFOUNDLAND

St. John's

CCMG Accreditation: No

Director: Dr. E. Ives, M.B., Ch.B., FCCMG, FRCPC

Maternal Serum AFP Studies: No laboratory services at The Dr. Charles A. Janeway Child Health Centre. No screening program but offered to high-risk cases, or to women requesting prenatal diagnosis for anxiety.

Computerized Data Base: No

Region Served: Newfoundland, Labrador, St. Pierre and Miquelon

Associated Laboratories:

Cytogenetics: Yes; part of Laboratory Services, Department of

Pathology, Janeway, director FCCMG.

Molecular: No
DNA Banking: Yes
Biochemical: No

Diagnostic Ultrasound: Mainly done at Janeway ultrasound

department, few at the Grace Hospital.

Outreach: Yes; Corner Brook, Grand Falls, Gander. Annually or bi-annually. Few prenatal patients, most come into St. John's. Amniocentesis may occasionally be done at Corner Brook and more rarely at Gander. Abnormal results phoned to obstetrician of record and to Genetics.

Comments: Small centre with limited range of services.

Appendix 4. Molecular Prenatal Diagnosis Available in 1990

MARITIMES

Halifax

In 1990 all molecular tests were done for carrier detection. Requests for prenatal diagnosis were referred out.

Molecular prenatal testing available in 1991:

Cystic fibrosis Duchenne type muscular dystrophy Phenylketonuria

QUEBEC

Laval

Myotonic dystrophy

University of Montreal

Duchenne type muscular dystrophy Cystic fibrosis

McGill

Thalassaemia Sickle cell anaemia Cystic fibrosis Tay-Sachs disease

ONTARIO

Ottawa

Myotonic dystrophy

Kingston

Haemophilia A Adrenoleucodystrophy Fragile X syndrome (linkage analysis only)

Additional molecular tests available in 1991:

Fragile X syndrome (by direct mutation)
Charcot-Marie-Tooth disease (X-linked)
Pelizaeus-Merzbacher disease

Hamilton

Haemoglobinopathies

Toronto

TORONTO PRENATAL DIAGNOSIS PROGRAM

The Hospital for Sick Children

Cystic fibrosis

Duchenne type muscular dystrophy

Wiskott-Aldrich syndrome

X-linked mental retardation

Neurofibromatosis

X-linked retinitis pigmentosa

Norrie's disease

VNTRs (Variable Number of Tandem Repeats)

Y probe

Chronic granulomatous disease

Choroideraemia

Carbamoyl-phosphate synthase

Ornithine transcarbamoylase

Polyposis

Autosomal dominant polycystic kidney disease

Additional molecular tests available in 1991: Spinal muscular atrophy Fragile X syndrome Retinoblastoma

North York

All requests for prenatal diagnosis were referred out. In the process of establishing a molecular laboratory in 1990.

ALBERTA

Calgary

Partial list of prenatally diagnosable disorders:

Haemophilia A
Muscular dystrophies
Alpha thalassaemia
Cystic fibrosis
Fragile X syndrome
Haemoglobinopathies
Huntington's disease
Neurofibromatosis
Molecular sexing
Lesch-Nyhan syndrome
X-linked disorders
Menkes' disease
Norrie's disease
Ornithine transcarbamoylase
Wiskott-Aldrich syndrome

BRITISH COLUMBIA

Vancouver

Cystic fibrosis
Duchenne type muscular dystrophy
Becker's muscular dystrophy
Myotonic dystrophy
Chronic granulomatous disease
Huntington's disease
Fetal sexing
Ornithine transcarbamoylase (OTC)

Additional tests available in 1991:

Alpha thalassaemia Zygosity testing

Appendix 5. Biochemical Prenatal Diagnosis Available in

MARITIMES

Halifax

GM₁ gangliosidosis Fabry's disease Tay-Sachs disease, I-cell disease Gaucher's disease Zellweger syndrome, peroxisomal disorders Niemann-Pick disease Metachromatic leukodystrophy

Biochemical tests available in 1991:

Developing very long chain fatty acid analysis for adrenoleukodystrophy and peroxisomal disorders.

QUEBEC

Laval

Active biochemical laboratory. Information on specific prenatal tests not supplied.

University of Montreal

Active biochemical laboratory. Information on specific prenatal tests not supplied.

McGill

Tay-Sachs disease

Haemoglobinopathies now rarely done by biochemical analysis.

ONTARIO

London

Comprehensive list not available.

Kingston

No information supplied.

Hamilton

McMaster does the testing for amino acid and organic acid disorders for much of Ontario.

Toronto

Toronto Prenatal Diagnosis Program

The Hospital for Sick Children

Gaucher's disease

Hurler's disease

Krabbe's disease

Metachromatic leukodystrophy

Tay-Sachs disease

GM, gangliosidosis

Pompe's disease

Pyruvate dehydrogenase

Pyruvate carboxylase

Succinate cytochrome c reductase

Cytochrome oxidase

Hunter's syndrome

I-cell disease

Sanfilippo's syndrome, type A

Sandhoff disease

NADH Coenzyme Q reductase deficiency

Additional molecular tests available in 1991:

galactosidosis

MELAS (Mitochondrial myopathy, encephalopathy, lactic acidosis and strokelike episodes)

MERRF (Myoclonus epilepsy associated with ragged red fibres) ATPase 6

MANITOBA

Winnipeg

All prenatal samples sent out for analysis in 1990.

SASKATCHEWAN

Saskatoon

Lysosomal hydrolases Galactosaemia Sandhoff disease

BRITISH COLUMBIA

Vancouver

No information supplied.

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An Assessment of the Readability of Patient Education Materials Used by Genetic Screening Clinics

Janis Wood Catano



Executive Summary

A number of factors, including reading level, writing style, and visual appeal, contribute to the readability of written materials. Also affecting readers' comprehension are the context in which they receive the material and the support provided to help them understand it.

This project reports on an analysis of the readability and comprehensibility of English-language patient education materials developed and/or distributed to clients by genetic screening clinics in Canada. Using the SMOG index and a resource evaluation checklist, 30 documents from 14 clinics across the country were assessed for content, writing style, organization, visual appeal, and illustrations.

Overall, the analysis found the material complex, technical, and difficult to read. However, a number of the documents also contained some positive aspects that could provide a basis for constructive change. In fact, several could serve as models for improvements to existing materials.

The study concludes by recommending that: (1) all existing materials and any new or revised materials be pre-tested with clinic patients as a part of development; and (2) genetic screening clinics

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jointly commission the development and testing of a readable patient information package.

Project Description

This project consisted of an analysis of the readability of Englishlanguage patient education materials developed and/or distributed to clients by genetic screening clinics in Canada.

Patient education materials from 14 genetic screening clinics were analyzed using the SMOG index and Catano and Breen's Resource Evaluation Checklist.¹

Readability Formulas

Essentially, readability formulas attempt to predict the level of difficulty of printed materials; that is, how easily the actual words and sentences in the text can be understood by the reader. A readability formula is a regression equation into which counts of selected language variables are inserted. Different formulas use different sets of variables, including, for example, the number of words per sentence, the number of sentences per 100 words, the number of one-syllable words, the number of polysyllabic words, and the frequency of occurrence of certain familiar words. The equation yields a score that is interpreted as an index of the readability of the material. More than 40 formulas are in common use, and recently various formulas have been developed and/or adapted for use in computer-based analysis.

The SMOG index was selected for the current analysis because it is widely used with health education materials, most notably public education materials on cancer. A review of readability formulas suitable for use with adult material written in the English language undertaken by the Office of Cancer Communications recommended the SMOG index on the grounds that it was "one of the simplest and fastest tests to use without sacrificing accuracy of prediction."²

The SMOG index assesses the reading level of a passage using a count of the number of polysyllabic words contained in a sample of 30 sentences. The number yielded by the SMOG grading formula is interpreted as the grade level of education necessary to ensure complete comprehension of the material. The SMOG index has a standard error of 1.5 grades.³

Resource Evaluation Checklist

Although the level of education required to understand the words of a text is one factor in its readability, it is not the only factor. The factors on which readability formulas are based can assess the reading level of a piece of printed material more or less accurately, but they do not cause the material to be readable. That is, writing materials with the aim of achieving

a designated score on a readability formula will not guarantee that they will be readable or that the reader will be able to comprehend the message. Doak et al. make this point very clearly:

It is worth making the distinction between the mechanics of reading and the process of understanding. To read — in the mechanical sense — we must simply have the skills to decode symbols that we call words and letters. But to understand what is read (decoded) we must also share with the writer an understanding of the logic implied in the passage, and we must have sufficient experience with the subject discussed and a working knowledge of the language used. For example, many highly literate professionals in other fields would be able to decode, but not understand, a research paper on astronomy or nuclear physics.⁴

Catano and Breen's *Resource Evaluation Checklist* was used to assess other factors influencing the readability and comprehensibility of the patient education materials.⁵ The checklist is a compilation of factors that past research has shown to influence the readability of printed materials.⁶ It offers a systematic means of looking at a variety of relevant factors, including the content, writing style, organization, visual appeal, and illustrations, and it enables evaluators to examine the material as a whole (see sample checklist in Appendix 3).

The Reader

Analysis of the readability of printed materials requires that one consider the intended reader. As has been pointed out previously, material that is readable and comprehensible to one group of readers may be unreadable, incomprehensible, or both, to another. A reader's understanding of what he or she reads is filtered through background, education, and life experience. For this reason, printed materials must be assessed in relation to the background, educational level, and experience of their intended readers. Assessors must try to see and understand the material through the eyes and experience of the intended reader, not through their own eyes and experience. It has been pointed out that health education materials far more often reflect "the reading ability of the designers rather than the potential target group."

The *Resource Evaluation Checklist* allows for consideration of several of these factors, including sex, age, ethnicity, educational background, economic status, and previous knowledge of a topic.

The primary intended readers of the material examined for this analysis are women who attend genetic screening clinics. A survey undertaken on behalf of the Royal Commission on New Reproductive Technologies provided the following details about the women who make up the readership.

Sex

All the respondents were female.

Marital Status

Although clients are sometimes considered to be couples, and are referred to as such in patient education material, not all of them were married. Fifty percent were currently married and living with their spouse, and 7.8% were in a common-law relationship. Of the remainder, 10.9% were single, never married; 17.2% were divorced; 12.5% were separated; and 1.6% were widowed.

Age

The respondents were older than the average childbearing woman — 88.5% were between 35 and 42 years of age.

Education

The respondents were well educated — 87.5% had completed high school and 56.2% had also completed some form of post-secondary education. Only 12.5% had less than a high school diploma.

Income

Respondents were relatively affluent — household income for the year was less than \$30 000 for 33.9%. It was between \$30 000 and \$59 999 for 45.8% and between \$60 000 and \$100 000 or more for 20.3%.

Employment

Most respondents were employed in skilled jobs. Employment was full time for 60.9% of the women, part time for 31.3%. Skilled jobs were held by 81.3%: for example, skilled clerical, sales, or service (23.4%); middle management (18.8%); semi-professional (15.6%); employed professional (14.1%); or high-level management (4.7%).

Ethnic Background

The respondents were ethnically varied. The largest group (23.0%) defined their ethnic background as Canadian. The second largest group (13.1%) was "other." Women identifying themselves as French Canadian accounted for 1.6% of this group. The other 20 ethnic identities ranged from 1.6% to 9.8%.

This information offers a description of persons who would be expected to read, try to comprehend, and try to make decisions based at least in part on printed material examined in the present analysis.

Method

Passages of educational material were obtained from 14 clinics across Canada. These items were evaluated by a single assessor who compiled the results from 10 January to 6 March 1992.

Results

Overall, the material was found to be complex, technical, and difficult to read.

In all, 30 items from 14 clinics were analyzed. Of these, 11 items received an overall rating of "poor," 15 "fair," 2 "good," and 2 "excellent" (Tables 1 and 2). The most frequent weaknesses were in the following areas:

- Reading level: The reading levels of the items ranged from Grade 11 to Grade 15: 2 were at the Grade 11 level, 7 at the Grade 12 level, 12 at the Grade 13 level, 6 at the Grade 14 level, and 2 at the Grade 15 level. Of the 30 items analyzed, one (a summary table) did not contain complete sentences and was, therefore, not suitable for analysis using a readability formula.
- Writing style: Eighteen of the 30 items were rated "poor," 9 "fair,"
 2 "good," and 1 "excellent."
- Visual appeal: Sixteen items were rated "poor," 10 "fair," 2 "good," and 2 "excellent."

A summary of the results of the readability analysis of each item, including comments and recommendations, is displayed in Appendix 1 (summary by source) and Appendix 2 (summary by diagnostic topic).

Discussion

When discussing the readability of printed materials, it is important to realize that, though components can be examined separately, they are interrelated and interdependent. The reader reacts to the document as a whole; moreover, this reaction occurs in a context that affects the reader's ability to read and understand the information.

The context in which the reader receives prenatal educational material and the supports that are offered to assist her to understand it have a powerful effect on her comprehension of the information she receives and her comfort with it. For example, several items suggested that the patient should read the material both before and after she meets with the doctor. Does she receive the information in the mail? Is it handed to her by a receptionist to read while she waits for an appointment? Some of the materials contain references to group or individual counselling. Is she given the material in a counselling session during which the material is used as a support or focus for discussion? All of these circumstances would affect the patient's reaction to the material and therefore her ability to read, understand, and act on the information.

Table 1. Resource Evaluation Checklist — Scores for 30 Items from 14 Clinics

			Categor
Score	Content	Writing style	Organization
Excellent	0	1	2
Good	30	2	3
Fair	0	9	12
Poor	0	18	13
Not applicable	0	0	0
Total	30*	30	30

For the purpose of this analysis, all materials were assigned the rating of "good" in th content category. This was done for two reasons: (1) because the content regardin the actual procedures was very similar for all clinics; and (2) because the evaluator ha no way of determining the accuracy of the information concerning procedures that wer specific to each clinic.

Table 2. Resource Evaluation Checklist — Summary of Results by Topic (n = 30)

				Categor
	Score	Content	Writing style	Organization
Prenatal diagnosis (n = 5)	Е	0	0	0
	G	5	0	0
	F	0	2	2
	Р	0	3	3
	n.a.	0	0	0
Chorionic villus sampling	Е	0	0	1
(CVS) and amniocentesis	G	8	1	1
(n = 8)	F	0	2	4
,	Р	0	5	2
	n.a.	0	0	0

^{**} One item (a summary sheet) did not contain complete sentences and was therefor unsuitable for analysis with SMOG.

			SMO	og
isual appeal	Illustrations	Overall assessment	Reading level	Number of items
2	2	2	Gr 11	2
2	. 0	2	Gr 12	7
10	6	15	Gr 13	12
16	8	11:	Gr 14	6
0	14	0	Gr 15	2
30	30	30		29**

1 fe			SMC	OG
isual appeal	Illustrations	Overall assessment	Reading level	Frequency
0	0	0	Gr 11	0
0	0	0	Gr 12	1
1	1	3	Gr 13	3
4	3	2	Gr 14	1
0	1	0	Gr 15	0
1	0	1	Gr 11	1
1	0	0	Gr 12	1
2	4	3.	Gr 13	2
4	1	4	Gr 14	1
0	3	0	Gr 15	2
			n.a.	1

Table 2. (cont'd)

				Catego
	Score	Content	Writing style	Organizatio
Amniocentesis (n = 6)	Е	0	1	1
,	G	6	1	0
	F	0	3	4
	Р	0	1	1
	n.a.	0	0	0
CVS (n = 1)	E	0	0	0
,	G	1	0	0
	F	0	0	0
	Р	0	1	1
	n.a.	0	0	0
Maternal serum alpha-	E	0	0	0
fetoprotein (MSAFP)	G	7	0	1
screening (n = 7)	F	0	2	2
,	Р	0	5	4
	n.a.	0	0	0
Down syndrome (n = 2)	Е	0	0	0
	G	2	0	1
	F	0	0 .	0
	Р	0	2	1
•	n.a.	0	0	0
Sickle cell anaemia and	Е	0	0	0
thalassaemia (n = 1)	G	1	0	0
,	F	0	0	0
	Р	0	1	1
	n.a.	0	0	0

Key: E = Excellent; G = Good; F = Fair; P = Poor; n.a. = Not applicable.

Well-thought-out support can do a great deal to improve the effectiveness of any kind of printed material. Although this factor is beyond the scope of the present analysis, individual clinics need to consider the effectiveness and readability of their materials in relation to the kinds of support they provide to their clients.

Reading Level

The reading level of the material was unacceptably high, despite the fact that the education level of the clinic patients was high compared to that of Canadians in general.

			SMC	OG
sual appeal	Illustrations	Overall assessment	Reading level	Frequency
1	1	1	Gr 11	1
0	0	1	Gr 12	1
3	1	4	Gr 13	3
2	1	0	Gr 14	1
0	3	0	Gr 15	0
0	0	0 .	Gr 11	0
0	0	0	Gr 12	0
0	0	0	Gr 13	0
1	1	1	Gr 14	1
0	0	0	Gr 15	0
0	1	0	Gr 11	0
1	0	1	Gr 12	2
3	0	4	Gr 13	4
3	1	2	Gr 14	1
0	5	0	Gr 15	0
0	0	. 0	Gr 11	0
0	0	0	Gr 12	1
0	0	1	Gr 13	0
2	0	1	Gr 14	1
0	2	0	Gr 15	. 0
0	0	0	Gr 11	0
0	0	0	Gr 12	1
1	0	0	Gr 13	0
0	1	1	Gr 14	0
0	0	0	Gr 15	0

The ability of a reader to absorb and use information is related to a variety of factors, including stress, anxiety, background knowledge, personal experience, and the type and extent of assistance available to support the printed materials. People have more difficulty in absorbing and understanding information when they are in an anxious or stressful situation. For many women, attending a genetic screening clinic would be stressful and anxiety-producing. Most would have little background knowledge about, or personal experience with, the procedures they might undergo. Therefore, reading difficulty would be expected to be exacerbated by anxiety.

Another reason to consider a lower reading level to be an advantage is that people tend to prefer material they perceive as easy to understand. In two different studies of pharmacy materials, patients found materials written at a Grade 5 level clearer and easier to understand than materials written at a higher level, regardless of their own reading abilities. People are more likely to read materials that they like.

A third consideration is that it is not unusual for people to have a reading level lower than their level of education. In a 1980 study, Doak and Doak tested the reading skills of patients in Virginia. Although most of the patients stated they were high school graduates, their word recognition skills were, on average, at about a Grade 7 level. Doak and Doak stated that "either their achievement had always fallen short of their grade placement, or else the skills they once possessed had diminished through disuse."

For these reasons, extra care needs to be taken to ensure that the reading level of patient education material is somewhat lower than the education levels of the clients.

It should be remembered when considering the reading levels of genetic screening educational material that the nature of the topic requires the use of many polysyllabic words. For example, the words *amniocentesis*, *prenatal*, *chorionic*, and *genetic* are all used frequently, and for the most part unavoidably, because part of the purpose of the patient education materials is to familiarize clients with the technical vocabulary.

Readability formulas have not been designed to take specialized health vocabulary into account. However, the SMOG formula is particularly effective with health materials precisely because it does highlight vocabulary and allow writers to identify problem areas, choose words and terms carefully, and develop ways to deal with specific issues. For example, it would be difficult to reduce the SMOG score on genetic education material much below Grade 11 without eliminating most of the technical vocabulary. However, this does not mean that the materials cannot be made more readable, or that once the essential technical vocabulary has been identified, efforts cannot be made to define terms clearly and to make all other parts of the document as uncomplicated as possible.

It should also be remembered that syllable length is not the only index of the difficulty of words. For example, the term *neural tube defect* contains no polysyllables, but that does not guarantee that readers will comprehend its meaning. In a study of unfamiliar words used in diabetes literature, Thrush and Lanese found that, though these topic-specific words accounted for only 19.6 % of unfamiliar words in the documents examined, they accounted for 66% of unfamiliar occurrences and contributed disproportionately to the reading difficulties of most of the materials. The authors suggested careful consideration should be given to minimizing the use of these kinds of words and, where it is crucial that the word be understood, that careful explanation be given. It is interesting that 99

(50%) of Thrush and Lanese's 198 unfamiliar health words were either one or two syllables in length.¹²

Although many of the materials examined in this analysis make some attempt to define new words in context, none of them contains a glossary or "Words You Should Know" list of any kind. Defining terms in a word list, as well as in context, would help to increase the reader's familiarity and comfort with the terms.

As noted previously, a score on a readability measure is only a rough guide to the difficulty a reader could be expected to have in understanding a piece of printed material. Tinkering with the text to attain a specific score on a formula will not make the material readable. However, using the readability score as an indication that the material might be difficult for the reader to understand allows the producers of the material to look at ways of improving the other factors affecting readability.

Writing Style

Many of the factors that would reduce the reading levels and increase readability are related to writing style.

The style in which the material is written has a profound effect on its readability. Writing style encompasses all aspects of the way in which the content is presented. Style includes point of view, tone of voice, and use of language. Style is the overall impression or feeling the material evokes in the reader. Style, in turn, is made up of many smaller factors, for example, the use of the active or passive voice, the use of concrete or abstract information, and the use of longer or shorter sentences. Style is intangible and can be difficult to describe, but its impact is clearly felt. Consider the difference in style in these two samples taken from materials from different clinics. They convey the same information but present it in markedly different styles.

The father will NOT be in the room with you.

We regret that, because of the large number of patients coming for amniocentesis, we cannot allow husbands to be present at the time of the procedure.

The writing style was a particular weakness in the materials analyzed for the present project. With few exceptions, the material was written in a very clinical style. That is, the style reflected the kind of technical, scientific writing with which most health and medical professionals are comfortable and familiar. It used the passive voice, dealt with facts rather than feelings, and was concerned primarily with transmitting information rather than experience. For example, most of the materials gave detailed, technical descriptions of amniocentesis. Relatively few described what the mother would experience.

There is no one style of writing that is appropriate for all circumstances or audiences. Although a clinical style is certainly effective

for communication between clinicians, it is not the style most appropriate for use in patient education materials. A more relaxed, informal style is easier for most people to read and understand.

The most direct approach to a less formal style is to write in a conversational tone — that is, to write as though the writer were speaking to the reader and the reader were someone whom the writer cared about. The use of a conversational style impacts on the readability of the material in several ways:

- Sentence length is more variable. Although short sentences are easier to read, material that is written using only short sentences can sound choppy, childish, and patronizing. Material that uses only long, complex sentences is difficult to follow and understand. Spoken sentences are usually shorter and less complex than written ones. The ebb and flow of conversation is also conducive to the use of many different kinds and lengths of sentences.
- The active voice is used more frequently. In a conversation, it is natural and easy to address a topic directly. The passive voice is less readable because it puts the subject closer to the end of the sentence. This means that the reader has to read the entire sentence to get to the point. For example, "The fluid is replaced by the body in about three hours" is passive; "Your body quickly replaces the fluid" is active.
- The tone is warmer and more personal. For example, patient education materials often refer to "the patient." It is difficult to imagine a conversation in which a clinician would address a client as "the patient." It would be natural to use the word "you." The use of "you" has the additional advantages of adding warmth and human interest to the material and enabling the reader to relate to the information. It also facilitates the process of presenting information from the patient's point of view, that is, focusing less on the details of the procedure and more on what the woman feels or experiences in relation to the procedure.

The following passages illustrate some of these points pertaining to writing style. Each of the passages in this first group of samples answers the same question concerning the accuracy of tests. These samples were selected because they range from a very technical, clinically detailed answer to a more direct, basic response.

SAMPLE 1A: Very technical information presented in a very clinical style

Chromosome analysis of cultured amniotic fluid cells is known to be highly accurate. Experience to date indicates that chromosome analysis of chorionic villus cells appears to be a reliable method for prenatal diagnosis. The cells taken at CVS come from the developing placenta and originate from the same fertilized egg as the embryo. The chorionic villus cells and the embryo are therefore assumed to contain the same genetic information. In the majority of cases, CVS appears to be as reliable as amniocentesis in detecting chromosomal abnormalities but various factors may affect the accuracy of both amniocentesis and CVS.

SAMPLE 1B: Much more direct, but still very clinical in content and tone

Chromosomal analysis of amniotic fluid and chorionic villi is highly reliable. Infrequently, a finding arises which may be difficult to interpret.

SAMPLE 1C: Very direct and basic, but still a bit stiff in tone, e.g., "is known to be" and "has also proven"

Amniocentesis is known to be very accurate. Chorionic villus sampling has also proven to be reliable, but there are more problems with interpreting the results.

SAMPLE 1D: Sample 1C, rewritten to be as direct and conversational as possible

Amniocentesis is very accurate. Chorionic villus sampling (CVS) is also reliable, but there may be more problems with understanding the results.

The four passages above illustrate the importance of considering the content in relation to what the reader wants or needs to know. The first passage contains information that may be primarily of interest to clinicians. It contains technical information, uses formal phrasing, and requires some background knowledge on the part of the reader. The subsequent passages are progressively less formal and provide direct information in a form that most readers would find easier to read and use.

The passages in the following set of samples refer to the need for the patient to drink water shortly before an ultrasound. Once again, the first passage is the most difficult. In these passages, the reader is asked to perform a specific activity — to drink before her ultrasound examination. In the first passage, this directive is buried in the middle. In the second it comes at the end. In the third it is stated early.

SAMPLE 2A: Required action placed in the middle of the passage, making it more difficult to identify

If you are scheduled for a CVS or an additional ultrasound scan either for dating of the pregnancy or a more detailed look at the fetus, it is necessary for you to drink at least 24 ounces of fluid prior to the examination. In these types of studies, it is important that the bladder be full in order to push the uterus up out of the pelvic area for a better visualization.

SAMPLE 2B: The result (a full bladder) emphasized, rather than the action (drinking water)

If you are scheduled for an ultrasound, **you must have a full bladder**. A full bladder will push the uterus up and provide a better image. It is important to drink at least three glasses of water prior to an ultrasound.

SAMPLE 2C: Above samples rewritten to emphasize the required action

Please drink at least three glasses of water shortly before your appointment for an ultrasound. When your bladder is full, it pushes the uterus up and allows us to see it more clearly during the examination.

The passages in the next set of samples show the influence of the point of view on the content of material. Different kinds of information become important depending on whose point of view is being considered. That is, does the material contain information that will tell the patient what she can expect to happen to her during an amniocentesis or does it give details about what the clinician does? Is the material written in a way that addresses the concerns of the patient directly, or does it deal with them indirectly, referring to the experience of some hypothetical "patient" rather than the reader herself?

SAMPLE 3A: Provides objective, clinical information, essentially unconnected to the patient

Amniocentesis is a procedure performed by an experienced obstetrician at approximately 15 to 16 weeks of pregnancy. A needle is inserted through the abdominal wall under ultrasound guidance into the water or amniotic fluid surrounding the fetus or baby. A small amount of fluid (less than ½ oz.) is removed and sent to a specific laboratory to be studied. The length of time to obtain results varies depending on the reason for the test being performed. Most results take approximately three to four weeks. No anaesthetic is used for the procedure and the majority of patients experience minimal discomfort.

SAMPLE 3B: Provides detailed information that describes what will happen, but uses the passive voice, refers to "the patient" or "the prospective parents," and makes no direct connection with the reader WHAT IS AMNIOCENTESIS?

A developing fetus is in a sac of fluid which contains cells that have been shed from the fetus. In amniocentesis, a needle is inserted into the uterus through the abdominal wall and a small amount (less than an ounce) of amniotic fluid is removed.

WHAT IS INVOLVED?

Before Testing

Testing is usually done at about 16 weeks of pregnancy (16 weeks from the first day of the last menstrual period). Prior to testing, the

family history is reviewed and risks and limitations of testing are discussed. The prenatal diagnosis clinic staff determine whether amniocentesis is indicated in each situation. Then the decision whether to proceed with testing is made by the prospective parents. Ultrasound Scan

Sound waves are transmitted by a special machine through the abdominal wall to form a picture of the fetus. This ultrasound picture shows the position of the placenta (or afterbirth), the development of the fetal head, spine and certain organs, and also whether there are twins present.

Amniocentesis Procedure

The amniocentesis itself takes about five minutes and is done in the Antepartum Assessment Unit of the hospital. It is suggested that patients rest in the waiting room for about 10-15 minutes following the procedure.

After Test

In a few cases, there may be slight cramping, spotting, or leakage of a small amount of fluid. If symptoms persist, the patient's doctor should be notified.

SAMPLE 3C: Provides information about what the patients will feel and in the final sentence uses "you" to make a direct contact with the reader

The actual procedure for obtaining a sample of amniotic fluid is performed by an experienced obstetrician and takes only a few minutes to complete. In preparation for the amniocentesis, an antiseptic solution is used to clean the abdomen. Most patients experience little, if any, discomfort. A pricking sensation, similar to a blood test, is felt as the needle enters the skin. Some patients describe the entry through the wall of the uterus as a sensation of pressure. A small amount of fluid (approximately 1 oz.) is removed for testing; this amount of fluid is usually replaced by the body within four hours after amniocentesis. You will be asked to remain for a short time afterwards for observation, and should make arrangements for someone to drive you home. It is preferable that you spend the remainder of the day at home engaging in quiet activities.

As these samples illustrate, the style in which the material is presented affects not only the readability of the material, but also the ability of the reader to relate that material to her experience and therefore to act on the information it provides. However, even before the reader encounters the content, she will have responded to the physical appearance of the material. The reader's response to the visual features of the material, including the production values and the illustrations, will be a factor in determining whether or not she reads it at all.

Visual Appeal

The visual appeal of printed material is made up of many details. These include the size of the type, the style of the type face, the length of the lines of type in relation to the type size, the colours of the paper and ink, the amount of type on each page, the way in which the blocks of type are arranged, the amount of empty space on the page, and the size and shape of the document. All these features contribute to an overall impression that influences the reader's perception as to whether or not the material is "readable." That is, to be read, not only must the material be readable, it must be seen by the reader to be readable.

Earlier in the present report, conversational writing style was described as material written as though the writer were speaking to the reader and the reader were someone whom the writer cared about. This approach is relevant to visual style as well. Not only is it important that the material sound as though the producers cared, it must also look as though the producers cared.

Visual appeal was another weak point in the material investigated for the study. The most common problems were: smudged, blurry type; an overall messy, careless appearance; crowded, dense-looking text; and justified rather than ragged right typesetting.

In a few cases, some care had been taken with production, for example, producing the material as a brochure on glossy paper. However, in several of these instances, the efforts were undermined by the use of extremely small type and by crowding too much information into a small space.

Most of the material was produced on $8\frac{1}{2}$ by 11 inch letter size paper. This seems to have been a matter of convenience, but it is not necessarily a problem. However, little effort was made to capitalize on the advantages, such as the large page size, offered by this format.

Most of the problems related to visual appeal can be easily and inexpensively corrected. The material produced by Chedoke-McMaster Hospitals provides a good model, using the letter-size page format to advantage by employing a large, easy-to-read typeface, ample white space, ragged right typesetting, and bold type for emphasis.

Conclusion and Recommendations

Although the material studied was generally difficult to read, many of the items had positive aspects that could provide the basis for a readable package. For example, much of the material was well organized and thorough. In many cases, relatively minor and inexpensive changes would result in a vast improvement in the readability of the material. In addition, there are several excellent models to offer guidance in making changes to existing materials, for example, "Patient Information: Prenatal Genetic

Diagnosis," produced by Chedoke-McMaster Hospitals, and "Amniocentesis and Ultrasound for Prenatal Diagnosis," produced by the Concern for Children Project of the Ontario Chapter of the Imperial Order of the Daughters of the Empire (IODE) and the Association of Genetic Counsellors of Ontario.

Recommendations

- 1. All existing materials and any new or revised materials should be pre-tested with clinic patients as a part of their development.
 - Pre-testing with the intended audience provides immediate feedback on the readability, clarity, credibility, usefulness, and consistency of the material as perceived by the people who will be using it. Pre-testing need not be elaborate or expensive, and it provides invaluable information that can be incorporated into the content and presentation of patient education materials.
- 2. Genetic screening clinics should jointly commission the development and testing of a readable patient information package.

Many of the items reviewed for this analysis are similar to one another and appear to have been developed based on materials produced by other clinics. Some items are direct copies, with only minor changes to reflect local policy. Such sharing indicates that a considerable amount of communication and sharing of resources among clinics already exists. By pooling their resources, a group of clinics would be able to afford to carefully develop the content, adequately test the resource, and produce a better quality of materials than would be possible for individual clinics.

Source of Material
Summary:
Checklist
Resource Evaluation
- :
Appendix

amniocentesis. Nice summary at end. Could be very good with more attention to style and presentation. Recommend use? Yes, with changes in style and format.	After a good introduction, this becomes very difficult to follow because of complex information, big words, not much guidance from the heads. Illustration is very badly placed in the text. (It illustrates the procedure for CVS, but is adjacent to text describing pregnancy termination.) The illustration is also difficult to interpret because it shows both cervical and abdominal CVS procedures. Recommend use? No. Use would require outside support.	Similar to the CVS material from the same clinic, but better produced and presented, especially the summary chart. Some parts
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	Information on Chorionic Villus Sampling	Information on Amniocentesis

who are not getting

	Writing Content style	Information on Amniocentesis (cont'd)	Prenatal G P Diagnosis	Summary of G F Amniocentesis and Chorionic Villus Sampling	A Comparison G P of CVS and Amniocentesis
Asse	Writing Organiza- Visual style tion appeal		۵	ш	ш
Assessment summary			۵	۵	O
ummary	Illustra- tions		۵	۵	n.a.
	Illustra- Reading tions level		Gr 14	е. С	Gr 15
	Overall		۵	ш	ш
	Comments	of the pamphlet have a nice tone (e.g., the use of "you" to personalize the material). The material itself, however, is very difficult and complex. The technical charts are very difficult to understand. Recommend use? Yes, with some revision and support.	Presentation is without warmth or empathy. Poor use of illustrations.	Good approach. Acceptable if used with support. Could be very effective with minor changes in layout and illustration. Recommend use? Yes, with support and changes.	Similar in content to the material of the same title

				Asse	Assessment summary	ummary			
Source	Title	Content	Writing style	Organiza- tion	Visual appeal	Illustra- tions	Reading level	Overall assessment	Comments
Halifax (Atlantic Research Centre for Mental Retarda- tion)	Maternal Serum Alpha- Fetoprotein Screening (MSAFP) (cont'd)						٦		paragraph 3. However, sentence length is good, with an effort to keep the material brief. The reassuring tag line at the bottom of the page is a nice touch. Recommend use? Yes, with support or rewrite.
	Maternal Serum Alpha- Fetoprotein Screening	o	۵	ш	۵.	n. Sa	Gr 13	ш	The major problem with this piece is poor visual appeal. It is blurry and smudged — very badly produced. This overshadows its assets: although the style is cold, the material is well organized and to the point. The reading level is better than most. This material could be easily improved with a minor rewrite and better production. Recommend use? Yes, with minor rewrite and better production.
Hamilton	Patient Information:	o i	G	ш	ш	n.a.	Gr 12	ш	Obviously, this is the product of much thought and effort.

it is beautifully presented; visually very clear and appealing, and well organized. The personalized front page is an asset. It is very clearly written, though style could be warmer (e.g., more use of "you" and less of "the mother"). Covers amniocentesis and CVS. Recommend use? Yes.	Obviously, this is the product of much thought, effort, and consultation. It has a warm tone, has human interest, and does not try to say too much. Question and answer format is effective. Cover is not as inviting as the rest of the pamphlet. Recommend use? Yes.	Content and tone are very technical. Question and answer format is effective, but more work is needed on style, tone, and clarity. Illustration is good but not well placed in the text. This pamphlet is almost identical to the one with the same name used by Ottawa, but
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Prenatal Genetic Diagnosis	Amniocentesis and Ultrasound for Prenatal Diagnosis	A Comparison of CVS and Amniocentesis
	Kingston (IODE and Assn. of Genetic Counsel- lors)	

Append	Appendix 1. (cont'd)								
				Asse	Assessment summary	ummary			
Source	Title	Content	Writing style	Organiza- tion	Visual	Illustra- tions	Reading level	Overall assessment	Comments
Kingston (IODE and Assn. of Genetic Counsel- lors)	A Comparison of CVS and Amniocentesis (cont'd)								the Kingston version is better printed and produced. Recommend use? Not as is, but it could be easily improved.
London	Testing in Pregnancy by Chorionic Villus Sampling or Amniocentesis	G	ш	C	ш	n.a.	Gr 13	ш	Question and answer is an effective format. Tone is a bit stiff, but better than some Information is well organized. Page 1 is crowded and dense. Line length is too long for easy reading. Recommend use? Yes. Minor revisions would improve it a great deal.
North York General Hospital	Alpha- Fetoprotein (AFP) in Pregnancy	Ø	۵	۵	ш	۵	Gr 12	۵	Brief and well printed, but the style is very impersonal and distancing. Instructions for completing form are unclear. Could be good with a rewrite.

				Asse	Assessment summary	ummary			
Source	Title	Content	Writing style	Organiza- tion	Visual	Illustra- tions	Reading	Overall	Comments
North York General Hospital (cont'd)	Sickle Cell and Thalassemia Screening (cont'd)								characters per line. Tone clinical and cold. Recommend use? Not without revision.
Oshawa	Amniocentesis Information Sheet for Patients Attending The Prenatal Diagnosis Clinic	C	U	ш	ш	ë -	Gr 12	o	This material has many good points, especially in regard to its writing style, which is much better than most. The use of "you" and the active voice makes it less formal and more human than most. The material is well organized and explains clinic procedures clearly and reassuringly. Visually acceptable, except for the overuse of boldface and capitals, which is distracting. Recommend use? Yes. It could be excellent with only minor revisions.
Ottawa	Spina Bifida	g	ш	O	O	ш	Gr 13	g	Much better writing style than most. The only real problems are small type size

explained. Subheadings are question format for headings is effective, but more work is Recommend use? No, not Cover illustration is excellent. A clearly non-white or ethnic crowded and dense and the good but not well placed in Recommend use? Would needed on style, tone, and warmer than most, but it is Content and tone are very Would be excellent with a clarity. The illustration is be acceptable with minor without a major overhaul clinical. It is visually very Recommend use? Yes. somewhat awkward and difficult to relate to main unpolished, as though it The writing style is a bit technical, and material were a draft. Using a child would be a plus. graph is not clearly seems somewhat few changes. evisions. the text. neads. م ш Gr 12 ш ட ۵. ۵ 0 م م ш G G A Comparison Amniocentesis (Is It For Me?) Birth Defects of CVS and Testing for Prenatal

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		Comments	Despite a high reading level, this is clearer than most and care has been taken to explain issues clearly. The tone is warmer than most. Recommend use? Yes, with minor revisions for style and reading level.	Brevity is an asset, but even so, the material looks crowded, dense, and uninviting. It is not clear to the reader at whom the information is aimed. It has a nice tone in some places (e.g., "We offer"), but is very clinical in others. Recommend use? Not without revisions.	This material is similar to that used by several other clinics and shares its strengths and weaknesses. It has a high reading level, a technical, chilly tone, and lacks warmth and human—
		Overall assessment	ш	۵	م
		Illustra- Reading tions level	Gr 14	£1 13	Gr 13
	summary	Illustra- tions	e,	ë L	ш
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	Asse	Writing Organiza- Visual style tion appeal	ш	۵	ш
		Writing style	ш	۵	<u>.</u>
		Content	G	ت ت	O
Appendix 1. (cont'd)		Title	Neural Tube Defects and Prenatal Diagnosis	Anencephaly	Information on Chorionic Villus Sampling/ Amniocentesis
Append		Source	Ottawa (cont'd)		Saska- toon

		Comments	Similar to material used by Toronto, North York, and Oshawa clinics. Recommend use? Yes, with a rewrite to reduce reading level and add some warmth.	Tone and style are more relaxed and warmer than most. However, this indication of caring is counteracted by the overall visual impression, which is very slapdash and messy. The material seems almost thrown together (e.g., the change of type size and style in the middle of the pamphlet). It is fairly well organized, has a question and answer format, but has too many cross-references. Illustrations are hard to see and interpret.
		Overall assessment		ц
		Illustra- Reading tions level	đ	Gr 13
	Assessment summary	Illustra- tions		Ф.
	essment	- Visual appeal		۵
	Ass	Writing Organiza- style tion		щ
		Writing style		ш
		Content		g
Appendix 1. (cont'd)		Title	Information Sheet for Patients Attending The Wellesley Antenatal Genetics Clinic (cont'd)	Information About Prenatal Diagnosis
Appendix		Source	Wellesley (Toronto) (cont'd)	Winnipeg

ic Topic
Diagnostic
Summary:
Checklist
Evaluation
Resource
Appendix 2.

	Comments	Presentation is without warmth or empathy. Poor use of illustrations. Recommend use? No.	Visually, pages 2 and 3 were good, but page 1 was poor — small type, crowded, and hard to read. The writing style is stiff, formal, and cold, though the material is well organized and comes in small units. Recommend use? Yes, with support or rewrite.
	Writing Organiza- Visual Illustra- Reading Overall style tion appeal tions level assessment	۵	ш
	Reading level	Gr 14	Gr 13
Assessment summary	Illustra- tions	۵	ю́.
ssment s	Visual appeal	<u> </u>	ட
Asse	Organiza- tion	۵	ட
	Writing style	۵	۵
	Writing Content style	ŋ	Q
	Title	Prenatal Prenatal diagnosis Diagnosis	Genetic Prenatal Diagnosis
	Topic Title	Prenatal diagnosis	

				Asse	Assessment summary	ummary			
Topic	Title	Writing Content style	Writing style	Writing Organiza- Visual style tion appeal	Visual appeal	Illustra- tions	Illustra- Reading tions level	Overall assessment	Comments
Prenatal diagnosis (cont'd)	Prenatal Testing for Birth Defects (Is It For Me?)	g	ш	<u>a</u>	۵	ш	Gr 12	ш	The writing style is a bit warmer than most, but it is still somewhat awkward and clinical. It is visually very crowded and dense and the graph is not clearly explained. Subheadings are difficult to relate to main heads. Recommend use? Would be acceptable with minor revisions.
	Information About Prenatal Diagnosis	U	щ	ш	<u>.</u>	Δ.	Gr 13	ш	Tone and style are more relaxed and warmer than most. However, this indication of caring is counteracted by the overall visual impression, which is very slapdash and messy. The material seems almost thrown together (e.g., the change of type size and style in the middle of the pamphlet). It is fairly well organized, has a question and answer format but has

too many cross-references. Illustrations are hard to see and interpret. Recommend use? Yes, with some design and editorial changes.	Poor on all counts. Difficult visually and with regard to content. Very technical vocabulary, with no definitions. Chart and graph require more explanation. Reads as though it was written for clinicians or medical students. Recommend use? No.	Good approach. Acceptable if used with support. Could be very effective with minor changes in the layout and illustration. Recommend use? Yes, with support and changes.	Similar in content to the material of the same title used in several other centres (e.g., Kingston and Ottawa). Although this version is much more visually appealing than most of the others, its reading level is
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	Untitled. Initial head reads: "Table 1: Indications for Genetic Prenatal Diagnosis"	Summary of Amniocentesis and Chorionic Villus Sampling	A Comparison of CVS and Amniocentesis
		Amnio- centesis and CVS	

Content style tion appeal tions level n G P F P F Gr 13 s s s	A Comparison of CVS and Amniocentesis (cont'd) Information on Chorionic Villus Sampling/Amniocentesis
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Content and tone are very technical. Question and answer format is effective, but more work is needed on style, tone, and clarity. Illustration is good but not well placed in the text. This pamphlet is almost identical to the one with the same name used by Ottawa, but the Kingston version is better printed and produced. Recommend use? Not as is, but it could be easily improved.	Question and answer is an effective format. Tone is a bit stiff, but better than some. Information is well organized. Page 1 is crowded and dense. Line length too long for easy reading. Recommend use? Yes. Minor revisions would improve it a great deal.	Content and tone are very technical, and material seems somewhat unpolished, as though it were a draft. Using a question format for headings is effective, but more work is
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A Comparison of CVS and Amniocentesis	Testing in Pregnancy by Chorionic Villus Sampling or Amniocentesis	A Comparison of CVS and Amniocentesis

penai	Appendix 2. (com d)			Asse	Assessment summary	ummary			
Topic	Title	Content	Writing style	Writing Organiza- Visual style tion appeal		Illustra- tions	Illustra- Reading tions level	Overall assessment	Comments
Amnio- centesis and CVS (cont'd)	A Comparison of CVS and Amniocentesis (cont'd)								needed on style, tone, and clarity. The illustration is good but not well placed in the text. Recommend use? No, not without a major overhaul.
	Patient Information: Prenatal Genetic Diagnosis	U	O	ш	ш	ё :	Gr 12	ш .	Obviously, this is the product of much thought and effort. It is beautifully presented, visually very clear and appealing, and well organized. The personalized front page is an asset. It is very clearly written, though style could be warmer (e.g., more use of "you" and less of "the mother"). Covers amniocentesis and CVS.
	Information on Chorionic Villus Sampling/ Amniocentesis	O	Д	ш	۵	ш	Gr 15	۵	This shares the strengths and weaknesses of other, similar materials: high reading level, technical, chilly tone, lack of warmth and human interest. However, it is well organized with a good

				Asse	ssment s	Assessment summary			
Topic	Title	Content	Writing style	Writing Organiza- style tion	Visual appeal	Illustra- tions	Reading level	Overall assessment	Comments
Amnio-centesis (cont'd)	Information Sheet for Patients Attending the Wellesley Antenatal Genetics Clinic	U	ш	LL	۵	.a.	Gr 14	ц	Overall appearance difficult to assess from a fax version. Good points are organization, introduction, and information on what patients might like to know about the procedures. Similar to material used by Toronto, North York, and Oshawa clinics. Recommend use? Yes, with a rewrite to reduce reading level and add some warmth.
	Information Sheet for Patients Attending the Antenatal Genetics Clinic	G	ш	ш	۵	ш	Gr 13	ш	Overall appearance is seriously compromised by the poor quality of the type. It looks blurry and is hard to read, which offsets the many good points of the presentation. It has nice detail about when to read the material, and good information about what the patient experiences during

Amniocentesis and ultrasound are covered. Recommend use? Yes, with a rewrite to reduce the reading level and add some warmth.	This material has many good points, especially in regard to its writing style, which is much better than most. The use of "you" and the active voice makes it less formal and more human than most. The material is well organized and explains clinic procedures clearly and reassuringly. Visually acceptable, except for the overuse of boldface and capitals, which is distracting. Recommend use? Yes. It could be excellent with only minor revisions.	Purpose is not clear. Tone is clinical and cold. Presentation is not bad, but length of lines (75 characters — 52 or less is optimal) makes the material more difficult to read. It has good information about what the patient can expect to experience.
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	Amniocentesis Information Sheet for Patients Attending the Prenatal Diagnosis Clinic	Information Sheet for Patients Attending North York Genetic Screening Clinic

				Asse	ssment s	Assessment summary			
Topic	Title	Writing Content style	Writing style	Writing Organiza- Visual style tion appeal	Visual appeal	Illustra- tions	Illustra- Reading tions level	Overall assessment	Comments
Amnio- centesis (cont'd)	Information Sheet for Patients Attending North York Genetic Screening Clinic (cont'd)								Recommend use? Yes, with rewrite to reduce reading level.
cvs	Information on Chorionic Villus Sampling	Ø	۵	۵	۵	Ф.	Gr 14	۵	After a good introduction, this becomes very difficult to follow because of complex information, big words, not much guidance from the heads. Illustration is very badly placed in the text. (It illustrates the procedure for CVS, but is adjacent to text describing pregnancy termination.) The illustration is also difficult to interpret because it shows both cervical and abdominal CVS procedures. Recommend use? No. Use would require outside

so, the material tooks crowded, dense, and uninviting. It is not clear to the reader at whom the information is aimed. It has a nice tone in some places (e.g., "We offer"), but is very clinical in others. Recommend use? Not without revisions.	Single sheet format is effective. Brevity is a plus. It needs more warmth and a clear statement of the intended audience. Recommend use? Yes, with minor revisions.	Good effort made to explain all aspects of what is done during screening. Not immediately apparent that this material is for women who are not getting amniocentesis. Nice summary at end. Could be very good with more attention to style and presentation. Recommend use? Yes, with changes in style and format.
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	Spina Bifida: A Neural Tube Defect	Syndrome Syndrome Screening

Appendix 2. (cont'd)		
	cont'd)	
- 1		

Appendix 3. Sample Resource Evaluation Checklist

PART 1: DESCRIPTION

• Title: _____ Producer: Produced in: □ Canada □ US □ International • Format: _____ • Length: _____ • Cost: _____ Brief Summary of Contents: PART 2: READER • Sex: _____ • Age: _____ • Ethnicity: _____ Educational background: ______ Economic status: • Previous knowledge of topic: Is this material: ☐ Need to know ☐ Nice to know ☐ For specialists PART 3: MATERIAL **AUDIO/VISUAL MATERIALS** PRINTED MATERIALS · Age or Type of Intended • Reading Level: Audience: ☐ Formula or assessment (Subjective assessment) method used: ☐ Predicted reading level: _____ Content: Content: ☐ Accurate ☐ Accurate ☐ Unbiased information ☐ Unbiased information □ Complete ☐ Complete ☐ Up-to-date ☐ Up-to-date ☐ Useful to audience □ Useful to audience

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•	Writing Style:	•	Verbal Style:
0000 0000	Purpose of material clear Relaxed, informal style Human interest Language appropriate to audience Clear definitions of new words or concepts Positive tone Concrete information Active voice Maximum 2 clauses per sentence Varied sentence length	000 0 0	Purpose of material clear Relaxed, informal style Human interest Language appropriate to audience Clear definitions of new words or concepts Positive tone Concrete information
•	Organization:	•	Organization:
000000	Manageable units of information Logical flow New material builds on old Important ideas repeated Information easy to find Clear topic headings Easy to use table of contents (if needed) Easy to use index (if needed)		Manageable units of information Logical flow New material builds on old Important ideas repeated Information easy to find Length appropriate for age of viewer
•	Visual Appeal:	•	Audio and Visual Quality:
	Appropriate type size Easy to read type face Few difficult type variations (italics, ALL CAPS) Type variations easy to read (boldface, underlining) Printing clear and unsmudged Paper and ink colours easy to see and read Paragraphs well spaced Margins and white space used Margins ragged right		Good sound quality Good voice quality Good visual quality Appropriate images Effective images Good integration of audio and visual components

•	Illustrations:			• Prese	ntation:	
0 000000	Appropriate to age Appropriate to eth ground Appropriate to soc Clear Accurate Up-to-date Well placed in text Charts/graphs/ta explained and idea Illustrations not u	nic back ial class bles clea ntified sed	arly	☐ Approground ☐ Approground ☐ Up-to ☐ Visua	nd opriate to s o-date als clearly e	ocial class
• •	Act I. Domination					
	PRINTED MATERIALS					AUDIO/VISUAL MATERIALS
		Poor	Fair	Good	Excellen	t
	Content Writing Style Organization Visual Appeal	0		0 0	0 0	Content Verbal Style Organization Audio and Visual Quality
	Illustrations Reading Level					Presentation
	(Grade) Overall Assessment					
С	omments:					
W	ould you recomme	nd using	g this n	naterial?		
A	ssessed by:			_ Date	:	

Notes

- 1. G.H. McLaughlin, "SMOG Grading A New Readability Formula," *Journal of Reading* 12 (1969): 639-46; and J.W. Catano and M.J. Breen, *Resource Evaluation Checklist* (Halifax: J.W. Catano and M.J. Breen, 1991).
- 2. U.S. National Cancer Institute, Office of Cancer Communications, Readability Testing in Cancer Communications: Methods, Examples and Resources for Improving the Readability of Cancer Messages and Materials (Washington, DC: Department of Health, Education, and Welfare, 1979), 3.
- 3. McLaughlin, "SMOG Grading A New Readability Formula."
- 4. C.C. Doak, L.G. Doak, and J.H. Root, *Teaching Patients with Low Literacy Skills* (Philadelphia: J.B. Lippincott, 1985), 60.
- 5. Catano and Breen, *Resource Evaluation Checklist*; see also J.W. Catano and M.J. Breen, "Developing Health Teaching Materials That People Can Read," *Literacy* 12 (Spring 1987): 23-30.
- 6. See, for example, D.B. Felker et al., Guidelines for Document Designers (Washington, DC: American Institutes for Research, 1981); Doak et al., Teaching Patients with Low Literacy Skills; M. Cutts and C. Maher, Writing Plain English: Why It Should Be Done, How It's Been Done, How You Can Do It (Whaley Bridge, Stockport: Plain English Campaign, 1980); Canada, Multiculturalism and Citizenship Canada, Plain Language: Clear and Simple (Ottawa: Canada Communication Group Publishing, 1991); and L. Hilts and B.J. Krilyk, W.R.I.T.E.: Write Readable Information To Educate (Hamilton: Chedoke-McMaster Hospitals and Hamilton Civic Hospital, Hamilton General Division, 1989).
- 7. A.S. Blinkhorn and J.M. Verity, "Assessment of the Readability of Dental Health Education Literature," *Community Dentistry and Oral Epidemiology* 7 (1979): 195-98.
- 8. Doak et al., Teaching Patients with Low Literacy Skills; and Hilts and Krilyk, W.R.I.T.E.
- 9. M.L. Eaton and R.L. Holloway, "Patient Comprehension of Written Drug Information," *American Journal of Hospital Pharmacy* 37 (1980): 240-43; and R.C. Adams et al., "Readability: Its Applicability to Education of Patients by Pharmacy," *Hospital Pharmacy* 14 (1979): 654-62.
- 10. L.G. Doak and C.C. Doak, "Patient Comprehension Profiles: Recent Findings and Strategies," *Patient Counseling and Health Education* 2 (3)(1980): 101-106. Cited in Doak et al., *Teaching Patients with Low Literacy Skills*.
- 11. Doak et al., Teaching Patients with Low Literacy Skills, 30.
- 12. R.S. Thrush and R.R. Lanese, "The Use of Printed Material in Diabetes Education," *Diabetes* 11 (1962): 132-37.
- 13. U.S. National Cancer Institute, Office of Cancer Communications, *Making Health Communications Programs Work: A Planner's Guide* (Rockville: Department of Health and Human Services, Public Health Service, 1989).

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- —. Readability Testing in Cancer Communications: Methods, Examples and Resources for Improving the Readability of Cancer Messages and Materials. Washington, DC: Department of Health, Education, and Welfare, 1979.





Canadian Physicians and Prenatal Diagnosis: Prudence and Ambivalence

Marc Renaud, Louise Bouchard, Jocelyn Bisson, Jean-François Labadie, Louis Dallaire, and Natalie Kishchuk



Executive Summary

This is a survey of Canadian physicians likely to send their patients for prenatal diagnosis (PND) — i.e., obstetricians, paediatricians, radiologists who carry out more than 100 obstetrical ultrasound scans a year, and a tiered sample of general practitioners (GPs) performing more than five deliveries a year. This study analyzed data collected from November 1989 to January 1990 for Quebec, and from October 1991 to January 1992 for the rest of Canada. For Canada as a whole, 3 072 physicians completed and returned the questionnaire, a response rate of 52%. Respondents were representative of this specific population in each Canadian province, each medical specialty, and both official languages. The Atlantic provinces were grouped together for purposes of data analyses, as were British Columbia, the Yukon, and Northwest Territories.

The questionnaire contained more than 200 questions and was extensively pretested in Quebec and in France. One section concerned respondents' demographic, sociocultural, and professional characteristics. The other questions related to physicians' attitudes regarding

This paper was completed for the Royal Commission on New Reproductive Technologies in August 1992.

(1) the utilization of prenatal diagnostic procedures; (2) the seriousness of various conditions, some of which can now be diagnosed *in utero*; and (3) the social choices that PND technology developments have brought about (in particular, the acceptability of abortion, the role of the physician in the decision whether to abort, and the appropriateness of greater government regulation in this area).

Use of Procedures

In Canada, there was a weak consensus¹ among physicians (around 60%) that the age of eligibility for amniocentesis should be maintained at 35 years. Most of those who opposed this norm said they would raise the age to above 35 years. There was no consensus on the age at which chorionic villus sampling (CVS) should be available; however, the tendency was also toward reducing access. Of all our respondents, GPs had the greatest reservations in this regard.

As was the case for amniocentesis, there was a weak consensus on the need to perform only one ultrasound scan per pregnancy, with GPs at the lower utilization end of the spectrum and specialists at the higher end. Here again, there was a remarkable range of opinion. In Manitoba and Alberta, nearly half the physicians (40%) did not consider it essential to order an ultrasound scan during pregnancy; in Quebec, only 4% of physicians shared this opinion. Quebec physicians tended to order two ultrasounds per pregnancy. Moreover, Quebec was the only province where the great majority of physicians (89% versus 60% for the rest of Canada) considered it acceptable to use ultrasound to screen for anomalies.

Most physicians opposed expanding access to amniocentesis under government health plans for any reason whatever (anxiety on the part of the expectant mother, freedom of choice, selecting the sex of the fetus). Contrary to geneticists' practice guidelines, many physicians (51%) said they did not feel justified in offering amniocentesis to a woman who would in any case refuse an abortion if an anomaly were diagnosed. They would be prepared, however, to expand access if women paid for the test themselves.

As regards new technological developments, physicians said they would be prepared to introduce predisposition testing for various common diseases, provided it were used in early childhood or during adulthood rather than prenatally. They accepted artificial insemination as a means of preventing the transmission of genetic disorders, but were much less sympathetic to surrogate motherhood. They were opposed to various procedures that would make it possible to select the sex of a fetus.

Multivariate analysis showed that physicians' attitudes toward procedures were less conditioned by cultural factors (religion, religious practice, ethnic origin, number of children) than social and professional characteristics. Apart from the influence of the province where they practised, the more direct contact they had with PND and the older they were the more they tended to favour technological development and the utilization of PND techniques.

Perception of Anomalies

Perceptions of the seriousness of the abnormalities listed in the questionnaire varied greatly. Generally speaking, anomalies resulting in a low degree of autonomy (paraplegia, trisomy 21 [Down syndrome], intellectual deficiency) were perceived as more serious than those suggesting future behavioural problems (e.g., aggressiveness) or fertility problems. But there were broad disparities among provinces. For instance, the majority of physicians in Quebec (70%) said they could not see themselves living with a child with trisomy 21, compared to a minority (40%) in the other provinces (and less than 20% in Saskatchewan). In addition, many more Quebec respondents (more than 84% as opposed to 61% for Canada as a whole) considered intellectual deficiency as serious.

Multivariate analyses showed that disparities in how seriousness is perceived are determined more by individual factors than by membership in a given group (i.e., sociocultural and professional characteristics). A small part of the variance (10%) was nevertheless attributable to practice area (urban or rural), religion, gender, number of children, province, specialty, and ethnic origin.

Social Choices

The Canadian medical profession unanimously and categorically rejected the use of PND for the purpose of selecting the sex of the fetus, just as it found unacceptable that a fetus of the "wrong" sex be aborted. Physicians rejected the idea of utilizing medical techniques for non-medical purposes.

Fifteen percent of Canadian physicians were opposed to abortion following diagnosis of an anomaly, no matter what the anomaly might be. This figure was surprising, since in our previous surveys (France and Quebec) the percentage of physicians unconditionally opposed to abortion was never more than 5%. The remaining 85% of physicians were distributed along a normal curve, ranging from mildly opposed to extremely sympathetic.

Given the historical prominence of trisomy 21 with regard to the development of amniocentesis, we expected that a majority of physicians, as in our previous surveys, would accept abortion for this anomaly. Only 50% of Canadian physicians were receptive to the possibility, with extremely pronounced regional disparities (ranging from 25% in Saskatchewan to 70% in Quebec). In this respect, Quebec's Anglophone physicians (the group that was by far the most open to selective abortion for all kinds of anomalies) constituted a distinct group within Canada.

The percentage of physicians favourable to abortion for the other listed anomalies was even lower than for trisomy 21. Generally speaking, religion, religious practice, specialty, ethnic origin, and province of practice (i.e., the sociocultural and professional characteristics of physicians) were the best predictors of abortion acceptability. These variables, plus the perceived seriousness of various problems, accounted for more than 30% of the variance (as much as 60% of the variance in some provinces).

Similarly, four out of five Canadian physicians objected to statements favouring elimination of anomalies mentioned in the questionnaire. The data suggest that the more general the practices of physicians, the more they are opposed to any form of control designed to eliminate anomalies.

No consensus exists in Canada on whether it should be left entirely to parents to decide on abortion (50% in favour, 36% against). There was a weak consensus, however, that physicians should sometimes intervene in the parents' decision, in particular to oppose abortion where anomalies are considered minor. A number of doctors (between 16% and 63%, depending on the item) considered it part of their role to offer direction with regard to the decision to abort. However, while they sometimes found it difficult to do so, all physicians felt an obligation to disclose all the information they have to the parents, with the exception of fetal sex (37% opposed disclosure of sex).

Lastly, 62% of physicians surveyed accepted existing regulations on the eligibility age for amniocentesis. Fifty-nine percent agreed there should be expanded access to PND, but only if patients make a direct financial contribution. Over 70% would not oppose the development of predisposition testing. On the other hand, they gave greater priority to funding preventive social programs (prevention of low birthweight, antismoking campaigns, etc.) than to the development of genetic screening technology.

Discussion

On the whole, Canadian physicians likely to order PND for their patients are prudent, reserved, even deeply divided about certain aspects of PND, the seriousness of anomalies, and the social choices offered by technological progress. This "family portrait" of the Canadian medical profession is in a way reassuring, for there is enough debate and diversity among physicians to suggest that the future evolution of PND will continue to reflect the diversity of values in Canadian society.

Some findings are puzzling, however. The extremely broad attitudinal disparities among provinces suggest that, when it comes to PND (access, abortion, directiveness), the question of whether a woman undergoes PND changes dramatically depending on where she lives in Canada. Indeed, multivariate analyses showed that, even discounting the effect of sociocultural and professional characteristics of physicians. the fact of practising medicine in a particular province has an impact on their overall attitudes. Four groups of provinces emerge: Saskatchewan; (2) the Atlantic provinces, Manitoba, and Alberta; (3) Ontario and British Columbia, reflecting the Canadian average: (4) Quebec, characterized by the presence of two medical communities, one English-speaking and the other French-speaking, both differing from physicians in the rest of Canada. Provincial "cultures," in the sociological sense of the term, seem to exist as far as PND is concerned. These cultures influence medical attitudes — and no doubt behaviours and therefore the fate of pregnant women.

Lastly, the assumption that PND is imperceptibly drawing us down a slippery slope, with extremely negative social consequences, is

examined in light of our findings and various other contextual factors that will influence the future development of PND.

Chapter 1. Overview of Issues

Prenatal diagnosis (PND), a medical procedure viewed as both a public health measure and a preventive tool, elicits high expectations and numerous concerns. This rapidly expanding field includes a number of techniques that have spread very quickly; their development has been characterized by a considerable degree of improvisation and unpredictability (Weill 1990). Since the mid-1970s, several scientific events and numerous government reports have emphasized the need to evaluate and structure these new medical/genetic methods, and a number of recommendations, principles, and guidelines have emerged as a result.²

These innovations have occurred in a cultural context where the attitude toward technology and its expansion has been more or less favourable. Personal standards and values have an influence on the utilization and spread of innovations, particularly when they involve matters of life and death, as is the case with PND. Where PND is concerned, it is important to understand the cultural factors influencing the behaviour of physicians, who are among the key players in its development. Several studies indicate that members of the medical profession play a vital role in the spread of PND (Lippman-Hand and Piper 1981; Bell et al. 1985; Dawe 1988; Nippert 1991; Reid 1991). It is they who control the delivery of health care services and who are the main source of referrals to those services. Their preference for certain styles of practice helps guide the development of PND procedures. Interacting with their patients, physicians play a crucial role in the utilization of this technology and the reproductive choices it offers.

Therefore, it seemed useful to conduct a sociological survey of those Canadian physicians most likely to have to decide whether or not to recommend PND for their patients — i.e., general practitioners (GPs) who perform deliveries, obstetrician-gynaecologists, paediatricians, and

radiologists who carry out obstetrical ultrasound scans.

In this chapter, before presenting the survey objectives, we will briefly review the ideological, scientific, ethical, and social context surrounding the development of PND. First we will look at the expectations and concerns it has raised, and then describe the various aspects of the social dynamic shaping its development. Lastly, we will present the survey objectives and questions.

Expectations Raised by PND

Broadly speaking, "PND" covers a series of medical services designed to prevent health problems in unborn children. It may include the

examination of the ergonomic aspects of the expectant mother's work environment, the search for metabolic diseases such as diabetes, and the screening of women for rubella at their first family planning visit. However, it is generally agreed that PND may be defined as a set of procedures for studying fetal symptomatology (ultrasound scanning, amniocentesis, chorionic villus sampling [CVS], etc.), thus enabling the detection of a growing number of pathological conditions *in utero*.

Approximately 3% of neonates are affected by anomalies or malformations whose causes are still largely unknown. Of the 5 000 singlegene disorders catalogued to date, 200 to 300 hereditary diseases or malformations are detectable in utero (McKusick 1990) using present knowledge and technology. In some cases, diagnosis reveals physical and intellectual abnormalities of varying severity, such as spina bifida, trisomy 21 (Down syndrome), cystic fibrosis, muscular dystrophy, and anomalies of the sex chromosomes (XYY, XXY, and XXX syndromes). In other cases, it detects late-onset conditions such as Huntington's disease. Finally, PND makes it possible to identify certain fetal characteristics, including sex. In the last 10 years, the field has advanced so rapidly that it is now possible to diagnose an ever-increasing number of genetic conditions. Until recently, the discoveries related to relatively rare diseases, but the 1990s promise to be a major turning point. Research is being directed toward understanding the genetic components of a host of complex diseases (cardiovascular disease, cancers, Alzheimer's disease, diabetes, manicdepressive psychosis, schizophrenia, and alcoholism). The development of technology to detect in utero predisposition to these diseases is even being considered. Several countries and vast financial resources have come together to carry out the "biological project of the century," the complete mapping of the human genome.³ In short, genetics is assuming an increasingly important role in elucidating and attacking disease.

PND is a direct offshoot of the remarkable progress made in medical imaging in the past 10 years and the revolutionary advances in genetics and molecular biology. These technologies hold out the momentous promise that medicine will be able — even more than it is today — to help parents give birth to "normal" babies and to avoid the birth of babies suffering from malformations or a variety of genetic disabilities.

The goals of PND, as presented by medical experts (Royal College of Physicians of London 1989), are to make it possible for women and couples at risk to give birth to normal children, encourage people at risk to reproduce, reduce anxiety about reproductive uncertainties, and ensure the best possible treatment of afflicted children through early diagnosis. In other words, PND could, in theory, provide better control over reproduction and allow us to make better-informed choices, thus broadening our freedom. From this point of view, PND appears to be a victory of sorts over fate and destiny.

Concerns About PND

Along with these expectations, the extension of genetic testing to a steadily growing number of people and diseases and the increasing commercialization of these biotechnologies have raised a host of issues that perplex even the genetics community (Council for Responsible Genetics 1990; Holtzman 1989; Mattei 1989). Issues of concern include the coercive use of tests; potential threats to personal freedom; possible ill effects in the form of stigmatization, intolerance, or discrimination; and the possible emergence of a new eugenics (Henifin et al. 1989; Retsinas 1991). Medicine may now be able to diagnose a number of malformations and deficiencies, but in most cases it cannot treat them. The available choice is thus to abort presumably malformed fetuses, or give birth to children with disabilities.

Many people fear that the use of PND will lead to the selection of the characteristics of unborn children (size, gender, eye colour, etc.). This fear is all the more justified since studies seem to show that the use of technology for such purposes is gaining wider acceptance among geneticists. Sorenson's 1972-1973 study in the United States showed that only 1% of geneticists were in favour of prescribing sex selection techniques (Sorenson 1976). In 1977, Fraser and Pressor reported evidence in Canada that 21% of geneticists were in favour of such techniques. By 1988, the percentages had risen to 62% of U.S. and 47% of Canadian geneticists (Wertz and Fletcher 1989b). This acceptance was based on respect for the patients' autonomy and did not necessarily mean that the geneticist was personally in favour of the practice.

Another consideration is that the increased tendency to view pregnancy in medical and technical terms has led to a distinction being made between fetus and mother, thus raising issues of rights and the potential for conflict between the two. Mother and fetus could become, in this context, legal adversaries. Childbirth could thus become "judicialized," as suggested by some recent U.S. trials dealing with so-called "wrongful life" or "wrongful birth" cases. These trials raise complex questions. Does the birth of a defective child mean that the child has suffered a wrong? What kind of life is worth living? How far does a physician's responsibility go?

Such considerations somewhat dampen the expectations raised by the development of PND. PND may, paradoxically, lead to a wider range of choices and yet limit the freedom to choose. We want to prevent malformations and disabilities, but there is concern that women may someday be forced to undergo PND through a sense of responsibility toward their unborn children. They may be made to feel guilty if they refuse an ultrasound examination or amniocentesis. Or, in certain countries they may be ostracized by agencies that refuse to pay for the treatment of malformed children who could have been aborted. In short, PND can have a liberating effect, but it can also impose even more constraints,

requirements, and social control. While we may be able to predict the short-term effects of a "discovery," it is much more difficult to foresee its long-range consequences.⁵

In recent decades, the experience of human reproduction has been profoundly transformed by technical mastery over procreation, the breakdown of the family, the mass entry of women into the workforce, and declining birth rates. In addition, health care systems all over the world are under great financial strain, which, in the medium term, might result in a rationing of services for women who, for instance, refuse an abortion. When prevention involves the state as much as the parents, some fear that having a baby in future years may become somewhat analogous to buying a car, with a number of trial runs being made before placing the order, and various quality control tests being performed during manufacture.

PND is today at the confluence of what could be called three revolutions: the "revolution" in molecular biology, the cultural "revolution" in childbearing as a result of the social changes mentioned above, and the potential "revolution" in the way government controls the expansion of health care services.

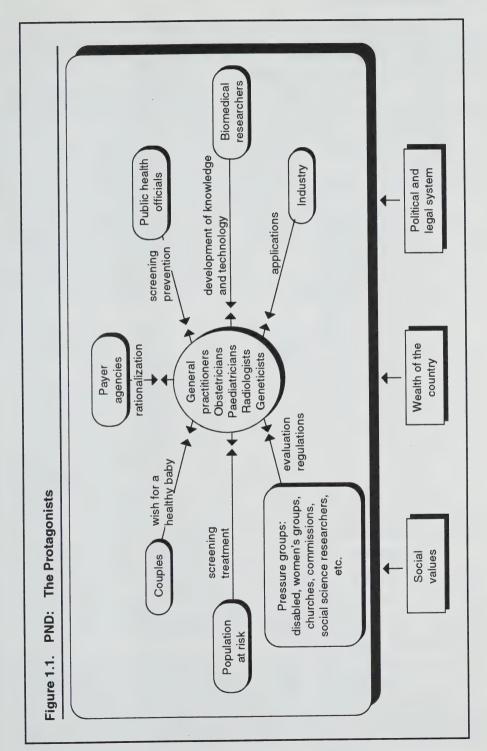
The conjunction of these "revolutions" gives PND an impact beyond that of most other medical innovations. Future developments in PND will depend on the attitudes of those involved, the social dynamics that evolve, and the regulatory mechanisms introduced.

Social Dynamics Surrounding Development of PND

We are witnessing an explosive increase in knowledge in the field of human genetics, an increase in knowledge that means an upheaval in social relations.

Medical practitioners are at the middle of the social dynamics surrounding the development of PND. Physicians (GPs caring for pregnant women, obstetricians, obstetrical radiologists, and paediatricians) use PND either as technical specialists or as attending physicians. In trying to make the best use of medical technology, they must satisfy the sometimes contradictory demands of parents who want both a beautiful baby (i.e., with the best medical guarantees) and a happy pregnancy (i.e., in the view of some, as technology-free as possible). In today's context, new babies are all the more precious because they are rare. Physicians must also take health care policies and costs into account. They have to deal with pressure groups, and they are targeted by the biotechnological industry and biomedical research circles, as Figure 1.1 illustrates.

PND practitioners are also caught in the middle of the moral and ideological debates that these technologies create. On the one hand, for instance, the Vatican condemns PND when carried out with the deliberate intention of aborting a defective fetus, while, on the other hand, geneticists (Wertz and Fletcher 1989b) and American obstetricians (Henifin et al. 1989) defend the reproductive freedom of couples and their freedom of choice with regard to PND and abortion. Lastly, for yet others (e.g., some doctors and



jurists), pregnant women who refuse PND knowing they are at risk of bearing a child afflicted with a serious anomaly should be brought to recognize their responsibilities. Such behaviour may be referred to as "prenatal abuse" (Henifin et al. 1989).

In short, to understand the trend of future PND developments, it is important to have an overview of the attitudes of those physicians whose practice includes the utilization of PND. Figure 1.2 illustrates what we believe to be the central role of the physician.

We shall examine the various elements of this diagram in turn.

Advances in Knowledge: Genetics as the Way of the Future

Since the turn of the century, a major change has occurred in infant mortality and morbidity rates. Improved living conditions have been a major factor in the decline of diseases linked to malnutrition and viral or bacterial infection. Hereditary diseases and congenital malformations have thus become the primary causes of infant morbidity and mortality, even though they affect only a small percentage of newborns. More than 50% of admissions to paediatric hospitals are for hereditary diseases, and more than 30% of deaths of children under the age of 15 are due to such diseases (Holtzman 1989).

Birth defects, as already pointed out, affect 3% of births. Statistics Canada reported 410 680 births in Canada in 1989; thus, there were potentially about 12 000 newborns with genetic anomalies.

Most birth defects are caused by unforeseeable errors in cell formation, a combination of genetic factors, disruptions in the very early stages of pregnancy, or complications during or after delivery. Causes of congenital malformations include such exogenous factors as infectious or toxic agents, ionizing radiation, and drugs (6.5%); endogenous causes are mutations, multifactorial heredity, and chromosomal anomalies (13.5%). However, the majority of embryopathies are of unknown etiology (80%) (Canada, Health and Welfare Canada 1988). In short, despite remarkable advances made in genetic knowledge, modern technology cannot eliminate all birth defects and it should not be expected to do so. It can simply allow certain high-risk couples to reproduce with greater confidence.

Genetic diagnosis is nevertheless perceived as the means of making an enormous contribution to our future knowledge of disease, its genetic and environmental origins, and its biochemical mechanisms. In particular, it will make it possible to detect predispositions to common diseases long before their actual onset. Medicine is thus on the point of becoming a predictive science, as well as a diagnostic and therapeutic one (Blumenthal and Zeckhauser 1989).

Geneticists at the leading edge of their discipline argue that the biological, internal aspect of disease contains an unknown component that can be understood through increased knowledge in the field of genetics. They believe that disease should no longer be seen as resulting only from outside aggression, but also as having an internal component. A paradigm shift appears to be required (Baird 1990). The concept of genetic

individuality and individual risk must be incorporated into disease prevention, changing the traditional way of looking at public health. This new idea of personalized prevention emphasizes the social gains that would result. Thanks to new technology, it will be possible to target individuals at risk instead of submitting a whole population to a prevention program.

Social Context Promoting Spread of Knowledge

During the past 20 years, we have witnessed major social changes with regard to reproduction: the drop in perinatal mortality and morbidity, greater mastery over procreation, a reduction in the number of children, more accessible abortion, changes in family structure, technological development, and unprecedented medicalization of the reproductive experience. Not only do doctors attend pregnant women, but the fetus itself has become a second "patient," viewed *in utero*. A new medical specialty has been born: fetal medicine. The social context, our new knowledge, and the resulting diagnostic possibilities raise expectations in some people for a trouble-free pregnancy and a healthy child.

For practitioners, these demands create dilemmas that the Canadian Medical Association (CMA) expresses in these terms:

It is somewhat ironic that the same advances in medical technology that have contributed to improvements in the quality of obstetrical care over the years have also induced an attitude on the part of some members of the public that an optimal medical outcome is to be expected in all cases. In the ensuing litigious atmosphere, a "normal or not unusual complication of treatment may be equated with negligence" ... This is a very real dilemma in the attempt to meet the psychosocial needs of the patient and the expectation of a perfect outcome and the technology it requires. (CMA 1987, 7)

Increasing Use of Procedures

PND consists mainly of procedures for visualizing the fetus (ultrasound screening, fetoscopy) and of sampling procedures (amniotic fluid, chorionic villi, and fetal blood). The former make it possible to study the morphology and structure of the fetus, the latter to carry out cell, chromosome, biochemical, and DNA analysis.

Several of these procedures are standard parts of sound medical practice today. Yet none of them existed prior to 1970: ultrasound has only been in use since 1974, amniocentesis since 1972, fetoscopy since 1976, and CVS and fetal blood and skin sampling since 1982. Obstetrical ultrasound is now used routinely during pregnancy (in 90% of pregnancies in Canada; in Quebec, there are 1.9 ultrasounds per pregnancy; in France, where use is reputedly the highest, 3.2).

Amniocentesis and CVS are aimed at certain groups of women, with age being the main criterion. Great efforts are being made to encourage women 35 years of age and over to undergo amniocentesis. In Canada, this group of women gave birth to 8.2% of children born in 1989. In 1990, 52% of them underwent amniocentesis, ranging from 64% in Quebec to 15% in

Newfoundland and 22% in Saskatchewan (Hamerton et al. 1993). In Quebec, the percentage reaches 80% for women 40 years of age and over (Dallaire 1991). More than 15 000 amniocenteses are performed annually in Canada. As with Canadian provinces, the rate of utilization of amniocentesis varies considerably from country to country. In Spain, less than 10% of women 35 years of age and over undergo amniocentesis, in Germany 50%, in France nearly 60%, and in Belgium 30% (Reid 1991).

CVS, on the other hand, is not as widespread a procedure (about 2 000 in Canada in 1990). Only 9.4% of women referred to a genetics centre undergo CVS. Studies on the risk of spontaneous abortion associated with CVS have yielded contradictory results. According to French data comparing series from around the world, the risk is as high as 5% (Boué 1989); according to a randomized Canadian study, it is comparable to that of amniocentesis (Lippman et al. 1992). Lastly, maternal blood sampling could someday be used extensively in all pregnancies, but remains controversial at this time. In Canada, Manitoba has experimented with it as part of a screening program, and it is used extensively in Ontario (Toronto and Oshawa) (Hamerton et al. 1993).

Proliferation of Ethical Issues

The rapid rate of innovation in medical genetics, the phenomenal increase in prenatal diagnostic alternatives, and the ever-expanding list of abnormalities entering into the decision whether to abort place genetics at the heart of the current social debate regarding the future of human reproduction.

The advent of PND has meant that genetics, instead of relying on statistics and probabilities as it did in the past, can now identify a larger number of disorders by looking directly into the womb. Yet it has no treatment to offer for several of those disorders. This situation results in the abortion dilemma following the diagnosis of genetic disease. Unlike abortion on demand, where pregnancy is unwanted, selective abortion occurs in the course of a pregnancy that is desired. What is the psychological impact on a woman of interrupting a desired pregnancy? What is the psychological impact of giving birth to a severely handicapped baby? Other moral issues emerge on a more collective level. Does PND carry the seeds of a new form of eugenics? Could PND redefine what is perceived as normal and abnormal? Is PND changing the threshold of tolerance toward imperfection and disability? In short, could PND be the catalyst for a new moral order?

The use of PND also raises issues concerning organization, information dissemination, and guidance — issues such as access conditions and criteria, access to services in a context of increased demand and financial constraints, definition of the populations that should receive the testing, orientation of prevention programs, and the use of genetic information.

Survey Objectives and Research Questions

The manner in which these technologies are integrated and disseminated depends on sociocultural contexts and value and representation systems, which vary among social groups, medical specialties, regions, and countries.

The general objective of this survey is to describe and explain the Canadian medical profession's attitude toward medical genetics, and more specifically toward the dissemination of prenatal diagnostic techniques and the numerous issues they raise.

The specific objectives fall into three groups:

- 1. learning about physicians' attitudes toward the use of technology, and their reasons for using it (when, why, and how a physician orders a particular PND procedure);
- 2. understanding the medical profession's attitudes toward various types of anomalies identifiable *in utero* and their opinions about the seriousness and severity of impairments and disabilities; and
- 3. understanding the medical profession's attitudes toward the broad social, ethical, and economic choices offered by the development of PND.

The simplest way to illustrate the general scheme of this survey is to describe its three main lines of inquiry (Figure 1.3). We will call them "Use of Procedures," "Perception of Anomalies," and "Social Choices." Each represents a specific objective of this study, and together they form the basis for the survey questionnaire (Appendix 1).

Use of Procedures

Questions relating to the "procedures" line measure the variables of the referral process with regard to PND: willingness to use PND, reasons for using it, opinion about the reliability of the procedure, opinion about the risk associated with it, and attitude toward patients who might be ambivalent or who refuse the test.

What interests us here is physicians' attitudes toward the implicit or explicit standards that guide medical practice, as well as their inclination toward expanding or reducing access to PND procedures.

The main techniques addressed by the survey are obstetrical ultrasound, amniocentesis, CVS, blood tests, and predisposition testing.

It should be recalled that obstetrical ultrasound is an imaging procedure used to obtain obstetrical data (age of pregnancy, position of the placenta, etc.) and for purposes of fetal evaluation. In the latter case, it makes it possible to probe not only external morphology, anencephaly, or malformation of the limbs, for instance, but also internal morphology that can indicate cerebral, cardiac, renal, or digestive tract malformations, or spina bifida. The study of fetal movements leads to a semiology of the development of the nervous system (Boué 1989). Ultrasound scanning has

been the subject of several studies dealing with the risks of the procedure, its effectiveness and diagnostic sensitivity, and its dissemination. It was also the subject of consensus conferences (United States in 1984, France in 1987). There is little indication that the use of ultrasound will decrease in future, although the number of ultrasound scans per pregnancy is still being debated, and its usefulness is sometimes questioned (Anderson and Allison 1990; Jacob 1986).

Fetal sampling procedures (amniocentesis, CVS, fetal blood sampling) are more invasive, but they add a new chapter to our knowledge of fetal development and the early diagnosis of genetic anomalies. Amniocentesis can be performed at different stages of pregnancy, but it is generally done around the sixteenth week. It is used mostly as a screening and diagnostic technique in women 35 years of age and older (38 in France), as such women are considered to have a higher risk for trisomy 21, a condition with an incidence in the population of 1/600 to 1/1 000 live births. In Canada, spontaneous abortion affects 0.5% of women who undergo amniocentesis. The skill of the person performing the procedure is vital to its success. Studies are in progress on early amniocentesis (i.e., around the twelfth week of pregnancy).

Sampling of the chorionic villi (the cells have the same genetic composition as the embryo's) is performed between the ninth and the eleventh week but, as previously noted, this test may possibly carry a higher risk. There is no doubt that the principle of a test capable of being carried out in the first trimester is well accepted by physicians and patients alike. Researchers are working to find a single test that would be the earliest, the most effective, and the safest.

The measurement of alpha-fetoproteins (AFP) in maternal blood is a screening procedure potentially applicable to all pregnancies. AFPs are proteins of fetal origin that are present in the amniotic fluid and in the maternal circulation. The level of AFPs in the blood can reveal the presence of neural tube defects, which are among the most common congenital malformations, and, more recently, of Down syndrome. The difficulties of interpretation, the diagnostic errors, and the ensuing investigations continue, however, to make it a very controversial screening test.

A few studies have dealt with the factors that influence the dissemination of these techniques. These include the level of knowledge of the procedures and a number of factors that are cultural (in particular, the attitudes of doctors and patients), financial, geographic (e.g., urban/rural disparity), and technical (e.g., laboratory skills) (Julian et al. 1986, 1989a; Nippert 1991; Reid 1991). The pressures exerted by various interest groups (pro-life groups, associations of the disabled, etc.) can influence the dissemination of techniques, as can the fear of lawsuits.

Dissemination raises a number of questions, the answers to which are subject to personal standards and values. For instance:

- Access to amniocentesis. The age criterion of 35 years as the 1. eligibility threshold for amniocentesis is to some extent arbitrary. This is the age at which the risk of spontaneous abortion roughly equals that of a trisomic birth. For all kinds of reasons (cost/benefit rationale, moral and ideological arguments, etc.), a number of people are uncomfortable with this criterion (U.S. President's Commission 1983; Crandall et al. 1986; Aymé et al. 1988; Moatti et al. 1990b), arguing in favour of either lowering the age or raising it. Should anxiety be considered a reason for expanding access? Must amniocentesis be reserved for cases where there is a presumption of serious and irreversible anomalies (Blancher and Frézal 1985; Fougeroux 1985; Maroteaux 1986; Wertz and Fletcher 1989b; Nippert 1991; Sjögren and Uddenberg 1990)? Given the costs and the risks of amniocentesis, should it be limited to women who would agree to abort in the event of a positive diagnosis (Society of Obstetricians and Gynaecologists of Canada [SOGC] 1983; Farrant 1985; Wertz and Fletcher 1989b)?
- 2. Access to ultrasound. Should ultrasound be made a mandatory component of pregnancy management? Should it be used less frequently? How reliable is it for detecting anomalies (Macquart-Moulin et al. 1989; Anderson and Allison 1990)? Should its use be expanded in order to reassure women or give them a sense of responsibility toward their fetus?
- 3. Role of the physician (directiveness). Is PND a matter of choice for women? Does the doctor have a right to strongly encourage a woman to undergo it? Can women have unconditional access to the tests if they pay (Wertz and Fletcher 1989b; Clarke 1991)?
- 4. Sex selection. Can these procedures be used for non-medical purposes, such as choosing the sex of the baby? Who has the power to decide, the parents or the professionals? Should information on the fetus's sex be withheld (Etzioni 1968; Fraser and Pressor 1977; Powledge and Fletcher 1979; Sorenson 1976; Ware 1987; Wertz and Fletcher 1989c; Reid 1991; Burke 1992)?
- 5. Reproductive methods for counteracting genetic disorders. To what extent is it acceptable to promote practices such as artificial insemination, surrogate motherhood, etc., to counteract genetic disorders (Royal College of Physicians of London 1989)?
- 6. The new genetics (predisposition testing) further complicates the issues raised by PND. There is concern that expanding genetic testing to cover a continually increasing number of people and conditions and the growing commercialization of the procedures could result in the coercive and discriminatory use of genetic testing, posing a threat to personal freedoms. Is there

a risk that genes will be manipulated for non-medical reasons (Lappé 1987; Holtzman 1989; Mattéi 1989; Fletcher 1989)?

These types of questions are examined in the present survey.

Perception of Anomalies

Although the manner in which physicians perceive disabilities is poorly documented, we thought it important to ask a certain number of questions so that we could study a possible link between their perceptions and acceptance of abortion.

The survey measures doctors' perceptions of the seriousness of various conditions such as paraplegia, intellectual impairment, sterility, and what is commonly called "behavioural problems." In the questionnaire, perceived seriousness is defined as the difficulty in living with a person afflicted with one of these conditions. In addition, we asked doctors to evaluate the gravity of certain syndromes.

We also examined their attitudes toward various anomalies diagnosable *in utero* and their acceptance of abortion. Several types of anomalies were selected in order to underscore the range of mental and physical impairments, depending on whether they threaten life at an early age, involve prolonged treatment, have an uncertain prognosis, or are lateonset. Those anomalies were the following:

- 1. Physical and mental impairments for which screening programs are in effect: trisomy 21; spina bifida; genetic diseases linked to populations at risk (muscular dystrophy, cystic fibrosis); Huntington's disease (an illness with an extremely gloomy prognosis, but which usually occurs in adults only), for which pilot screening programs are in effect for populations carrying this dominant genetic disorder; and phenylketonuria (a deficiency that can result in mental retardation, but can be controlled with a very strict diet), which can be diagnosed prenatally.
- 2. Malformations detected by ultrasound scanning, a procedure that is now an integral part of pregnancy monitoring: anencephaly, hydrocephaly, and facial, cardiac, diaphragmatic, abdominal, and skeletal anomalies. Some are fatal (anencephaly), while others are operable at birth, although they require several operations and these may not always be successful (e.g., hydrocephaly, cardiac malformations); a few are curable (e.g., certain digestive tract malformations, cleft lip and palate); and lastly, some cannot be corrected and result in physical disability (e.g., absence of a limb).
- 3. Anomalies of the sex chromosomes. These anomalies are not screened, but are revealed by chromosome study. Their prognosis is uncertain (XYY, Klinefelter's syndrome [XXY], Turner's syndrome [XO], and triple X syndromes). In some cases, slight deviations from the norm are observed in carriers. In other

cases, these anomalies involve problems with sexual development and sterility.

- 4. Predisposition testing. Some diseases can be linked to a genetic marker. They include insulin-dependent diabetes, mental disorders (some types of schizophrenia, manic-depressive illness), coronary heart disease, and alcoholism in some families. In theory, these tests make it possible to establish that there is a higher probability of certain diseases, but cannot predict whether the disease will, in fact, develop. These diagnostic tests can be expected to progress further.
- 5. Sex selection. Sex can be determined with certainty through amniocentesis and CVS. The earlier detection occurs, the more crucial the question of sex selection becomes.

Various factors influence the perception of anomalies, their seriousness, and the need to prevent them. Religious affiliation and practice, ethnic origin, even the physician's specialty can all have an influence. We shall discuss these factors as the survey's data are presented.

Social Choices

Again, the goal of this survey is to account for the cultural factors that shape the development of PND and to attempt to predict, based on today's attitudes and behaviour, what the future holds for PND and the application of newly gained knowledge in human genetics. We must therefore understand physicians' attitudes toward the procedures, anomalies, and the many social choices resulting from the wider use of these techniques and the more frequent diagnosis of possible abnormalities.

Thus, it seemed important to ask doctors about the social issues surrounding PND, the most significant of which are the following:

- 1. Regulation. Should access to the various prenatal diagnostic techniques be regulated even more than is currently the case? Or, on the contrary, should access to these procedures be a matter of supply and demand?
- 2. Abortion. Should we allow malformed fetuses to be aborted? Are new social standards of tolerance toward imperfection and abnormality emerging? Is there an implicit threshold beyond which abortion becomes socially and ethically unacceptable? Where do we draw the line? Should selective abortion be allowed for some abnormalities, and prohibited for others?
- 3. *The physician's role (directiveness)*. Should doctors have a say in such complex decisions? To what extent should doctors be absolutely neutral? Is neutrality possible or desirable?
- 4. *Information disclosure.* To what extent should doctors be required to disclose all information in their possession, even in

the case of minor anomalies or ambiguous data? Should they disclose the sex of the fetus without medical reason?

- 5. Funding priorities. Do our governments devote too much (or not enough) money to genetic services? Does our society attach too much (or not enough) importance to genetic, as opposed to social, disabilities (associated, for instance, with poverty, the very young age of the mother, or poor living habits)?
- 6. Harmful effects of PND. The development of PND raises many concerns for the future. Will information from these tests be used for discriminatory purposes? Will PND transform conditions hitherto considered normal into new disorders? Is there a danger we will end up with coercive discriminatory policies (compulsory abortion, laws to limit the transmission of harmful genes, etc.)?

There is an abundant literature on each of these questions and the social choices they present. We shall summarize only those studies dealing with the acceptability of abortion. Abortion is undoubtedly the most important of all these topics. Some regard it as the most vehemently debated issue of the late twentieth century (Kunins and Rosenfield 1991). What is more, PND lends abortion a special character. By freeing reproduction from biological chance and making it possible to interrupt pregnancy selectively, PND may be setting new social rules. Some fear it could help reduce the importance attached to the treatment of genetically disabled children and the resources allocated to easing their integration into family and community. Others disagree, believing that people's values and common sense will prevail, resulting in the appropriate use of these technologies. In short, this issue is so important that it warrants taking a closer look at the findings of previous surveys.

Research on Abortion Acceptability

Numerous American and European surveys have been conducted since the mid-1970s on physicians' and women's opinions and attitudes regarding PND. Tables 1.1 to 1.5 summarize the major findings of studies that have measured acceptance of selective abortion by doctors and women.

We will use a consensus approach to present the findings, as defined below. As can be seen in Table 1.1, when doctors were asked if they agreed with abortion for fetal anomalies, the results suggested a strong consensus (more than 75% in favour). A weak to moderate consensus (60%-75%) in favour of abortion has been developing since at least the 1980s. Except for the Quebec/France study, surveys indicate that only about 15% of doctors are now unconditionally opposed to abortion.

Studies on women with regard to PND indicate a moderate to strong consensus in favour of selective abortion (Table 1.2). Faden and colleagues' (1987) study shows that, if a fetal anomaly were diagnosed, slightly fewer women would consider an abortion for themselves (65% of women not in a

Table 1.1. Acceptability of Selective Abortion and Abortion on Demand Among Doctors, According to Various Studies (%)

	Agree with abortion (fetal anomalies)	Agree with abortion on demand	Abortion rejected regardless of circumstances
Carlos and Cloutier (1976) Quebec 2 500 doctors	78	27	14
Canada, Committee (1977) Canada 3 133 obstgyn. & GPs	82	22	17
American College of Obstetricians and Gynecologists (1985) United States 1 300 obstgyn.	91	75	13
Savage and Francome (1989) Great Britain 396 gynaecologists		73	
Renaud et al. (1993) Quebec/France 746 doctors (QC) ¹ 588 doctors (FR) ¹	71 ² 74 ²	57 63	5 ³ 4 ³

¹ GPs, obstetrician-gynaecologists, paediatricians, and obstetrical radiologists.

² Rate of agreement with aborting a fetus with trisomy 21.

screening program, 74% of those in a screening program for neural tube defects) than for others (81% and 86%).

Breslau's (1987) study is interesting in that the attitudes of mothers with children suffering from anomalies are comparable to those of mothers of normal children: about two-thirds of them accept abortion. However, when mothers with afflicted children were asked if they would abort a fetus with the same anomaly as one of their children, they were much more hesitant. Thus, only 25% of mothers with a trisomic child and 20% of those with a child suffering from cystic fibrosis (Wertz et al. 1991) said they would choose abortion, although they thought that selective abortion should be legal (see Table 1.3).

In Canada, public opinion polls show that one Canadian in four (24%) favours legalizing abortion on demand, while 60% would accept abortion "in

Percentage of doctors unequivocally rejecting abortion for 13 different conditions.

Table 1.2. Acceptability of Selective Abortion Among Women Concerned by PND, According to Various Studies (%)

Abortion justified for anomalies	Abortion considered for anomalies	Abortion rejected
77		
	71	6
	70	
	84	7
	89	11
99 84 83		
81	65	
86	74	24
65 68 79		
	99 84 83 81 86 65 68	justified for anomalies 77 71 70 84 89 99 84 83 81 65 86 74

Table 1.2. (cont'd)			
	Abortion justified for anomalies	Abortion considered for anomalies	Abortion rejected
Sjögren and Uddenberg (1988) Sweden		60 (aprilain)	
211 women (admitted for		62 (certain)	

93

35 (probable)

2

Moatti et al. (1988) France

amniocentesis)

353 women (admitted for amniocentesis)

·

Table 1.3.	Acceptability of	of Selective	Abortion	Among	Parents	with
an Afflicte	d Child (%)					

	Would choose abortion	Consider abortion should be legal
Hsia et al. (1979) Hawaii 167 parents of children with spina bifida		65
Elkins (1986) United States 40 mothers of children with trisomy 21	25	
Wertz (1991) United States 271 parents of children with cystic fibrosis (CF)	20 (CF) 35 (MMR*)	75 (for CF) 80 (MMR*)

^{*} Moderate mental retardation (MMR), as defined here, implies the ability to communicate, but not to attain self-sufficiency.

some circumstances" (*La Presse* 1991; *Toronto Star* 1991). Percentages are apparently higher in the United States, with 70%-80% for abortion on demand (Henshaw and Martire 1982; Munday et al. 1989) and more than 80% for selective abortion (various polls of the National Opinion Research Center; Granberg and Granberg 1980; Mulvihill et al. 1989).

As Table 1.4 shows, when doctors consider specific anomalies, they are much less open to abortion than when asked if they accept selective abortion in general. There was a moderate to strong consensus on trisomy 21 only. Abortion for cystic fibrosis and sex chromosome abnormalities is either highly controversial (50% agreement) or rejected (less than 30% in favour).

Finally, as can be seen from Table 1.5, long-term American, French, and Quebec studies indicate that more than 90% of pregnancies diagnosed with chromosomal abnormalities (including trisomy 21) are terminated. It should be pointed out that women who agree to amniocentesis envisage abortion in the event of a positive diagnosis. On the other hand, abortion rates for diagnosed sex chromosome abnormalities range from 41% to 63%.

To summarize, selective abortion — as a general concept — is better accepted than abortion on demand. On the other hand, there is a much wider variation when the abnormalities are specified. Hypothetically speaking, the percentage of persons in favour of abortion increases in proportion to the perceived clinical seriousness of the anomaly. It should be noted that doctors and female patients seem to have very similar attitudes.

Some of these studies also examined the factors influencing doctors' acceptance of abortion: religion, religious practice, age, medical specialty, gender, parental status, and cultural background.

Weisman et al. (1987) found that female doctors are generally more open to abortion regardless of the specific indication, but they are less inclined to use prenatal screening procedures. This is confirmed by other studies (Carlos and Cloutier 1976; American College of Obstetricians and Gynecologists [ACOG] 1985). On the other hand, when asked about abortion for fetal anomalies, they appear somewhat less accepting than their male counterparts. Younger practitioners seem more open to PND and abortion than their seniors (Carlos and Cloutier 1976; Weitz 1979; Bernhardt and Bannerman 1982; ACOG 1985; Julian et al. 1989b). Religious affiliation is also a very significant factor: Jews are most in favour, followed by Protestants, and then Catholics (Carlos and Cloutier 1976; Weitz 1979; Bernhardt and Bannerman 1982). Religious practice is a more consistent indicator than religious affiliation (Weitz 1979), and the fact of having few or no children also seems to have a bearing on abortion acceptance (ibid.).

Differences between medical specialties have been documented in the United States. Obstetrician-gynaecologists appear more sympathetic to abortion than do paediatricians and GPs (Weitz 1979). Paediatricians are considerably more pessimistic than psychologists, educators, health professionals, and social workers regarding the prognosis of mental retardation (Wolraich and Siperstein 1986). The doctor's relationship with a patient is also a factor: the better the rapport, the more open the doctor appears to be to abortion (Nathanson and Becker 1977, 1981).

Carlos and Cloutier (1976) also observed differences among medical specialties in Quebec, but the difference between Anglophones and

Table 1.4. Acceptability of Selective Abortion Among Doctors for Various Anomalies, According to Various Studies (%)

Authors	Trisomy 21	Turner's syndrome	Trisomy Turner's Klinefelter's Spina 21 syndrome syndrome bifida	Spina bifida	Cystic	Cystic Tay-Sachs fibrosis disease	Thalassaemia Haemophilia	Haemophilia
Weitz (1979) United States 445 doctors¹	65	47				74		
Margolin (1978) United States 350 social science/ genetics students	8				54		52	24
Julian et al. (1989b) France 853 doctors¹	78	47	44	52	46			21
Renaud et al. (1993) Quebec/France 746 MDs (QC) ² 588 MDs (FR) ²	71	29 27	20	38 55	46 64			

1 GSs, obstetrician-gynaecologists, and paediatricians.

GPs, obstetrician-gynaecologists, paediatricians, and obstetrical radiologists.

	Autosomal trisomies	Sex chromosome abnormalities
Golbus et al. (1979) United States 2 978 women (amniocenteses)	(58 out of 59) 98	(6 out of 10) 60
Benn et al. (1985) United States 7 000 women	(57 out of 59) 97	(18 out of 29) 62
Verp et al. (1988) United States 4 684 women	(42 out of 48) 88	(14 out of 20) 41
Holmes-Siedle et al. (1987) GB-Finland 7 299 women		(25 out of 40) 63
Baird et al. (1985) British Columbia 2 957 women	(47 out of 47) 100	(16 out of 16) 100
Briard (1990) France 63 362 women		(91 out of 174) 52
Dallaire et al. (1991) Quebec 19 790 women	(318 out of 340) 94	(37 out of 60) 62

Francophones is the most striking. According to their study, 80% of Anglophones would agree to abortion for various conditions, compared to 54% of Francophones.

As for women, the type of advice they receive has an influence on whether they decide to abort (Julian et al. 1989b). Being advised by an obstetrician-gynaecologist is more likely to lead to a decision to abort than being advised by a geneticist (Verp et al. 1988). Participating in a screening program apparently results in a greater acceptance of abortion (Faden et al. 1987). Early diagnosis (CVS as opposed to amniocentesis) could become a determining factor in the future (Verp et al. 1988).

Outline of Report

Following a description of survey methodology (Chapter 2), the report is divided into two parts. The first section (Chapters 3-6) contains the

overall findings for the three main lines of inquiry explained above. Chapter 3 details the sociocultural and professional profile of the physicians surveyed. Chapter 4 describes the main similarities and disparities, province by province. Chapter 5 does the same for each of the four medical specialties, while Chapter 6 looks at physicians' attitudes in relation to their religious affiliation/practice. The second section (Chapters 7-8) contains the multivariate analyses covering all the variables that could be predictive of physicians' attitudes. The statistical model employed was multiple classification analysis. Chapter 7 contains the principal findings and Chapter 8, a description of the general model that emerges from them. Significant geographical subcultures were evident, indicating that attitudes toward PND vary considerably across Canada. The conclusion provides a summary of the survey and guidelines for public policy on PND.

Chapter 2. Methodology

Previous Surveys in Quebec and France

This survey was based on a similar study conducted in Quebec and France in 1989 and 1990. The Quebec survey sample comprised 1 193 physicians who refer patients for PND. Of that number (998 Francophone and 195 Anglophone), 746 (63%) responded after three reminders. In France, 881 (58%) of the 1 473 questionnaires were completed and returned after two reminders. The Quebec data in this study came from this first survey. We have disregarded the French study (Renaud et al. 1991, 1992) for the purposes of this report.

Questionnaire

Quebec/France Version

The questionnaire was initially developed after a series of meetings with various Quebec and French experts and a systematic review of all comparable surveys conducted around the world on the ethical and social issues involved in PND. About 10 versions of the questionnaire were prepared, each being extensively discussed by the research team and doctoral students in sociology.

The questionnaire was designed to obtain a concise picture of physicians' attitudes and conduct with regard to the three lines of inquiry described above. In addition to general questions (attitudes, perception of standards, etc.), we tried, through case histories and scenarios, to determine the position physicians would adopt when specific problems of this nature were encountered in actual practice. The scenarios enabled us to understand the interaction between the three research areas. For most questions, physicians were asked to indicate where they stood on a five-point Likert scale ranging from "totally disagree" to "totally agree."

The question on social issues consists of a series of statements concerning the discriminatory or coercive use of PND, the importance attached to social as opposed to genetic disabilities, the physician's directiveness, his or her inclination to resort to the techniques at an early stage, liberal attitudes, and so on. The wording of this question was extensively reviewed for validity and reliability. A pilot survey was conducted on 115 fourth-year medical students at the University of Montreal and 62 first- and second-year family medicine residents.

Lastly, the entire questionnaire was pretested to ensure that questions would not appear biased, important questions would not be left out, all words would be understood, all questions would be interpreted in the same way by all respondents, the questionnaire would create a positive impression, and people would feel motivated to complete it. The final questionnaire was then pretested on some 50 physicians and other professionals from the University of Montreal, McGill University, University of Amiens, and University of Picardy.

An English version of the questionnaire was also developed and

retranslated into French to ensure the correct words were used.

The questionnaire used in Quebec and France consisted of 31 questions, including 10 on the practice of PND and the criteria for using it and expanding access to it (53 subquestions), two questions on the perception of seriousness of 13 conditions, one on the acceptability of abortion for 13 different fetal conditions, one question on the social issues generated by PND (36 statements), and 17 questions on professional and sociocultural matters (65 subquestions).

Canadian Version

The Canadian version of the questionnaire incorporated most of the Quebec/France version, but was slightly altered to suit Canadian conditions (see Appendix 1). In addition to the sociocultural questions that we reworked or added, we improved the wording of some questions. Three ethical problems in the Quebec/France questionnaire were deleted because they were not applicable, and four questions (Q14, Q27, Q28, and Q29) and one ethical problem (Dilemma 34) were added (Q14 was added at the suggestion of one of the Commission's working groups).

The new questions were translated into English and then retranslated into French for validation purposes. A professional graphic artist reformatted the questionnaire, and 11 500 copies were printed (French and

English).

Study Population and Sample

Study Population Defined

The study population was that of Canadian physicians likely to use PND and thus refer their patients for the procedure: GPs performing five or more deliveries per year, obstetrician-gynaecologists in active obstetrical practice, paediatricians, and radiologists performing 100 or more obstetrical ultrasounds a year.

In Quebec, we were able to draw up a list of the physicians meeting our criteria by cross-referencing the lists of the various professional federations concerned¹⁰ with data from the Régie de l'assurance-maladie du Québec (RAMQ), the Quebec health insurance plan. Since Quebec physicians have to be registered with the federations in order to practise, the lists are very reliable. Preparing lists in the other Canadian provinces was more difficult.

The CMA provided lists for Canada (excluding Quebec) compiled from the records of the provincial medical federations. However, there were problems with two of the specialties under study — GPs and radiologists.

We were unable to obtain a list of the total population of Canadian GPs performing five or more deliveries a year and therefore had to estimate it based on answers to a 1991 CMA survey. That survey covered *all* Canadian physicians and, at the time of our survey, 65% of Canadian physicians (43% of GPs) had responded. Physicians were asked to state the number of deliveries they had performed in the previous year. From the answers given, we were then able to draw up a partial list of GPs performing five or more deliveries a year and hence estimate their population in Canada.

Once we knew the percentage of GPs in the CMA survey performing five or more deliveries a year, we were able to estimate their population in Canada by multiplying the known total number of Canadian GPs by this percentage. We assumed that the CMA percentage of GPs performing five or more deliveries a year was representative of the actual percentage in the Canadian GP population as a whole. Since 43% of Canadian GPs answered the questionnaire, we believe this assumption was justified. To refine our estimate further, we calculated the percentage of these same GPs for each province and both language groups. Table 2.1 shows the total number of GPs, GPs performing five or more deliveries according to the CMA survey, the percentage of GPs performing at least five deliveries by province and by language group, and the estimated number of GPs carrying out five or more deliveries in the Canadian population.

The CMA was unable to provide us with the number and list of Canadian radiologists who perform 100 or more obstetrical ultrasounds a year, but did give us the number and a list of all Canadian radiologists. We therefore sent the questionnaire to all Canadian radiologists, asking those who performed 100 or more obstetrical ultrasounds a year to complete it. We had to estimate the population of these radiologists, and did so on the basis of the response rate of the other specialists (obstetricians and paediatricians). We did not use the GP response rate because it was higher than that of the specialists. As response rates were much the same for each specialty and within each province, we postulated that they would also apply for radiologists and based the latter's population on these rates. As can be seen from Table 2.2, the population of radiologists who perform 100

Franco. 749 23 Anglo. 7 274 33 187 94 2 308 1 045 2 036 459 401 Estimate Table 2.1. Estimated Population of GPs Performing More Than Five Deliveries per Year % 5 deliv. Franco. 9.86 0.20 0.25 0.05 4.07 % 5 deliv. Anglo. 23.34 53.80 43.52 55.36 48.53 36.67 32.88 1.32 84.62 29.4 N 5 deliv. Franco. 241 CMA survey respondents N 5 deliv. Anglo. 248 467 1 018 22 97 29 071 3 427 147 CMA survey N resp. 478 295 4 585 073 839 9 2 201 407 11 671 461 9 882 2 401 3 678 90 569 7 097 853 27 348 983 1111 z Population Oue. Ont. Man. Sask. Alb. B.C. N.W.T. Nfd. N.S. P.E.I. N.B.

Francophone	Estimated N 3 151 2	% response 40.0 48.1	Number of respondents 1 96 1 0	X 4.751	Estimated N 22 38 4 39 20 361 27 40 101	* response 44.6 49.5 48.1 47.3 44.8 44.4 41.6	0 0	Anglophone Number of respondents % resp 10 44.6 19 49.5 3 - 18 18 - 9 174 48.1 13 47.3 18 44.8 45 41.6
Number of response	1	ŧ	-	-				
Number of response % response 4 1 40.0 151 96 - 7 1 48.1 3 0 - 1 1 0 - 1 1 1 1 1 1 1 1 1 1 1 1 1 1	1	,	-	-	})	
Number of Number of Nesponse)	-	183		41.6	
Number of response % response 4 1 40.0 151 96 - 7 1 48.1 3 0			0	-	9 101		44.8 44.4	
Number of respondents % response 4 1 40.0 151 96 - 7 1 48.1	•	•	0	က	27		47.3	
Number of respondents % response 4 1 40.0	7	48.1	-	7	361		48.1	
Number of respondents % response	151	,	96	151	20		4	
Number of respondents % response	က	40.0	-	4	39			
Number of respondents % response					4			
Number of response					38		49.5	
Number of respondents % response					22		44.6	10 44.6
		% response		Z	imated N	Est	% response Est	% response

or more ultrasounds a year, by province and language group, was obtained by dividing the number of radiologists who answered the questionnaire by the assumed response rate. We thus estimated that 991 (835 Anglophone, 156 Francophone) radiologists in Canada perform 100 or more obstetrical ultrasounds a year. (It should be noted that, in Table 2.2, there is no need to *estimate* response percentages for provinces where none is indicated. In Quebec, for instance, the number of radiologists performing at least 100 ultrasound scans a year was already known.)

Sampling

The sample was tiered on the basis of language, province, and specialty: the language in which the physician received literature from the various medical associations or bodies, ¹¹ the province in which a physician was practising at the time of the survey, and the specialty in which a physician was registered with the various medical associations and bodies.

The sampling fraction varied with the various tiers. In Quebec, all radiologists, Anglophone GPs, and obstetricians, as well as two-thirds of

Francophone GPs and paediatricians, were selected.

Outside Quebec, all radiologists, paediatricians, and obstetricians were selected. The sampling percentage for GPs varied from province to province to ensure a sufficient number of physicians was included for each province.

Some Respondents Excluded

Once the questionnaires were received, some respondents had to be eliminated from the sample because they did not fit the definition of the population under study. These included physicians who were retired or performed purely administrative duties, radiologists carrying out fewer than 100 obstetrical ultrasounds a year, obstetrician-gynaecologists not practising obstetrics, and GPs with fewer than five deliveries a year. 12

Summary

The number of Canadian physicians sampled outside Quebec is as follows:

Physicians on CMA lists 9 000
Less Quebec physicians (based on CMA lists) 1 977
Total physicians in population 7 023
Physicians not selected at time of sampling 1 743
Physicians sampled 5 280
Less respondents excluded
Less radiologists not meeting criteria
Total number of Canadian physicians sampled
outside Quebec

The figure of 4 760 is used in the weighting procedures described later.

Data Collection

Survey Protocol

Physicians are a difficult professional group to survey. Their heavy workload, the large amount of paperwork they have to do each day, and their perplexed attitude toward social research may explain why they show little inclination to answer questionnaires.

In order to obtain the best possible response, we employed the following techniques, which, according to the relevant literature, ¹³ have proved to be the most successful:

- the questionnaire and mailing/return envelopes were designed and printed by a professional graphic artist;
- the questionnaire was accompanied by a letter signed by the Chairperson of the Royal Commission on New Reproductive Technologies and the President of the CMA;¹⁴
- the questionnaire was also accompanied by a letter from the researchers, explaining survey objectives;
- advertising appeared in the medical press during the survey period (see *Canadian Medical Association Journal* 145 (November 1991)); and
- two reminders were mailed.

All correspondence with physicians was carried on in their language of choice. For the survey outside Quebec, the first mailing went out on October 7, 1991, the first reminder on December 2, and the second reminder on December 18.

Response Rate

Of the 4 760 physicians in the non-Quebec sample, 2 334 completed and returned the questionnaire (49%). When Quebec data were combined with those for the rest of Canada, the response rate was 51.6% (3 072 respondents out of 5 953 questionnaires mailed). Table 2.3 shows the number of respondents, the response rate, and the weighted number for each tier.

Building the Data Base

Coding

A coding system was used for written answers, those left blank, and those that did not coincide with the choices given or allowed. Appendix 2 shows the coding for "religion" (validated by the Canadian Centre for Ecumenism) and "ethnic origin." Some answers in the Quebec sample had to be recoded to coincide with the choice of answers offered in the pan-Canadian questionnaire.

5.38 8.56 5.38 11.50 8.07 0.00 0.98 152.87 116.67 Total Weighted no. of resp. 10.52 65.30 Respondents, Response Rates, and Weighting, by Language, Specialty, and Province % resp. Francophone No. of resp. Survey basis z Weighted 1.47 0.00 0.98 5.38 8.56 11.50 15.41 resp. 116.67 no. of 50.00 0.00 51.06 44.44 50.00 % resp. 40.91 45.71 Anglophone No. of resp. 0000 38 24 28 19 13 9 01 Survey basis 22 9 8 4 62 47 63 38 22 22 22 22 22 477 47 63 38 9 8 4 22 35 22 22 33 Z Prince Edward Newfoundland Paediatricians **Paediatricians** Paediatricians Obstetricians Obstetricians Nova Scotia Obstetricians practitioners practitioners Radiologists Radiologists practitioners Radiologists Table 2.3. General General General Island

			Anglophone	ne				Francophone	hone		
	z	Survey basis	No. of resp.	% resp.	Weighted no. of resp.	z	Survey basis	No. of resp.	% resp.	Weighted no. of resp.	Total
New Brunswick General											74.85
practitioners	187	59	16	55.17	45.74	23	12	œ	66.67	5.63	51.37
Obstetricians	27	27	10	37.04	09.9	4	4	2	50.00	0.98	7.58
Paediatricians	22	22	10	45.45	5.38	2	2	2	100.00	0.49	5.87
Radiologists	39	39	8	46.51	9.54	7	7	***	20.00	0.49	10.03
Quebec General											474.73
practitioners	94	25	5 5	88.00	22.99	700	327	226	69.11	171.21	194.20
Paediatricians	166	69	36	56.52	40.60	233 411	227	124	54.63	100.52	141.12
Radiologists	20	20	თ	45.00	4.89	151	151	96	63.58	36.93	41.82
Ontario General											999.15
practitioners	2 308	694	423	60.95	564.50	19	6	4	44.44	4.65	569.15
Obstetricians	621	621	315	50.72	151.89	12	12	10	83.33	2.94	154.83
Paediatricians	756	756	319	42.20	184.91	9	9	က	50.00	1.47	186.38
Radiologists	361	361	174	48.20	88.30	7	7	-	20.00	0.49	88.79
Manitoba General											152.35
practitioners	401	87	51	58.62	98.08	-	-	-	100.00	0.24	98.30
Obstetricians	64	64	26	40.63	15.65	-	-	0	0.00	0.00	15.65
Paedialricians	130	30	90	46.15	31.80						31.80

l	112.26 10.03 11.74 9.78	353.17 256.08 29.35 43.04 24.70		498.47 38.89 50.62 44.76	6.83	6.11 0.24 0.24 0.24	5.14 0.024 0.024	3 072
143.81	0.24	0.00	632.74	0.49		0.24		399.39
	100.00	0.00		100.00 66.67 100.00		100.00		63.14
	-	0 +		- 01 -		-		899
	-	-		- m -		-		1 058
ı	-			0 e -		-		635
	112.26 10.03 11.50 9.78	256.08 29.35 42.80 24.70		497.98 38.16 50.38 44.76		6.11 0.24 0.24	5.14 0.24 0.24	2 672.57 1 635
	62.60 48.78 40.43 45.00	50.98 45.00 42.86 44.55		56.19 47.44 39.98 41.53		54.55 100.00 100.00	75.00 100.00 100.00	49.11
ľ	30 20 19	78 54 75 45		109 74 70 76		9	o	2 404
	48 41 47 40	153 120 175 101		194 156 206 183		====	5	4 895
ľ	459 41 47 40	1 047 120 175	m.	2 036 156 206 183		25	2	10 930
Saskatchewan	General practitioners Obstetricians Paediatricians Radiologists	Alberta General General practitioners Obstetricians Paediatricians Radiologists	British Columbia	practitioners Obstetricians Paediatricians Radiologists	Northwest Territories	practitioners Obstetricians Paediatricians Radiologists	Yukon General practitioners Obstetricians Paediatricians Radiologists	Total

Weighting

The data were entered and verified. We estimated the data entry error rate at 1 character in 6 500. We then combined the Quebec and Canadian data and determined the weightings.

The weight of each sample tier was determined using the following equation:

 $W^2 = I^2 \times R$

where

W² = theoretical weight of each respondent

I² = inverse of the sampling fraction for each group of physicians

R = inverse of the response rate fraction

We therefore established a tiered weighting by specialty, province, and language so that the percentages of physicians in the various tiers would be the same as for the population concerned. It should be pointed out that this weighting alters the percentages of physicians in the various tiers, while leaving the size of the sample unchanged. Data are accurate to ± 1.80 for the physician population as a whole, with a confidence level of 95%.

To prevent any methodological bias stemming from the use of different sources to determine a population, we decided to use data from a single source — the CMA — to establish the population in each tier, even though CMA estimates differ somewhat from those of the Corporation professionnelle des médecins du Québec. 15

Analytical Methods

Our data analysis method comprised three steps: (1) we analyzed the distribution of Canadian physicians' answers to each question and the main areas of agreement and disagreement by province, specialty, and other sociocultural variables; (2) we created and validated modelling scales; and (3) we conducted multivariate analyses of those scales.

Canadian physicians' answers to the questionnaire and the main cleavages are cross-tabulated. Each table gives the percentage of physicians (by province, specialty, or other variable) who answered a given question in a particular way. The answers on the Likert scales were recoded so as to have only three types of answers: for, against (depending on the context, it could be "agree" or "disagree"), and a mid-point answer between these two opposites. Categories 1 and 2 on the five-point Likert scale were combined for this purpose, as were 4 and 5. The relative percentages of physicians for or against a particular position are thus easier to compare. This process also made it possible to condense the information and clearly identify those questions on which there was consensus. The chi-square test made it possible to determine whether the percentages for the various answer categories varied significantly from one group to the next. However, as this test was significant for nearly all cross-

tabulations because of the size of the sample, we used adjusted standardized residuals to identify the widest differences. The residuals indicate the degree of disparity between the percentage observed and the percentage that might be expected if there were no difference between the groups. As the residuals are normally distributed with a mean of 0 and a standard deviation of 1, 2.6 or more indicates significant differences with an alpha threshold (probability of error) of 0.01.

A table summarizing areas of consensus follows the analysis of the main cleavages by province. This will make it easier to understand the main issues and points of agreement in Canadian physicians' practice and opinions regarding PND. Similar tables also follow the sections on medical

specialties and religion.

In order to summarize survey results, and follow up on the multivariate analyses conducted as part of the Quebec/France survey and subsequent modelling, we have reproduced and validated various scales. with reference to acceptability of abortion, the perception of the difficulty posed by various anomalies, directiveness of the physician with regard to the decision whether to abort, and expanded access to amniocentesis and Each scale was created by combining the answers to various questions measuring a single concept. When compared to the individual answers, these scales have the advantage of providing a stable estimate of an attitude or general concept. They also have the advantage of being interval scales and therefore more amenable to multivariate analysis, which allows all important factors to be evaluated simultaneously. Reliability tests, notably Cronbach's alphas (Cronbach 1951), make us confident that the scales are internally consistent and statistically valid.

The multivariate analyses consist of mostly variance analyses (F tests) and multiple classification analyses (MCA) (Andrews et al. 1967). Variance analysis makes it possible to identify variables that have a significant effect on the scales, and MCA allows evaluation of the direction and intensity of these effects. Thus, it is possible to check whether a physician's specialty has an influence on his or her acceptance of abortion and, if so, which specialty has a greater or lesser acceptance. It should be noted that MCA is particularly well suited to the use of non-independent categorical predictors, a designation that applies to most of the variables in this survey

(e.g., medical specialties, provinces, religions, etc.).

Part 1. Outcomes

Chapter 3. Sociocultural and Professional Profile of Physicians

In this chapter, we present the sociocultural and professional profile of physicians who answered the questionnaire. The data show the distribution of physicians using PND, by specialty, for Canada as a whole, and for each province. We will present sociocultural and professional data, yielding a profile of the physicians and their practices. The use made of PND by the various specialties is also described. To make the text easier to read, only the most informative tables and figures are included. Detailed statistical tables appear in Appendix 3. Table references are indicated in parentheses in the text. This procedure is followed in subsequent chapters.

Quebec's Medical Structure Is Different

The study population consisted of GPs performing five or more deliveries a year (n = 1 962, weighted data), obstetricians (n = 373), paediatricians (n = 495), and radiologists who carry out 100 or more obstetrical ultrasound scans a year (n = 242) across Canada (n = 3 072). The findings are presented by province, except for the Atlantic provinces (ATL/Atl. in tables and figures), which are combined because of the small number of physicians who practise there, and the Northwest Territories and the Yukon, which are combined with British Columbia (BC/NWT). Tables 3.1 and 3.2 describe the study population, the sample, and response rates, by specialty and by province.

There were approximately twice as many GPs as specialists in the sample as a whole: 1 962 GPs compared to 1 110 specialists (obstetriciangynaecologists, radiologists, and paediatricians), as Figure 3.1 shows. Quebec differs dramatically from the rest of Canada, with far fewer GPs than specialists (194 GPs and 281 specialists). GPs therefore form the vast majority of physicians using PND in Canada. In Quebec, a number of GPs have abandoned obstetrics, while the number of specialists is, on the whole, comparable to that of other provinces. This affects data interpretation in two ways:

- (a) PND is dominated by more specialized physicians in Quebec. This caused one of our consultants to remark: "You won't understand your findings unless you know that Toronto is a city of general practitioners, while Montreal, like Boston, is a city of specialists. As for the rest of Canada, PND is clearly dominated by GPs, with a few specialists in the large cities."
- (b) This difference explains the disparities observed in the number of physicians in the sample compared with the total population

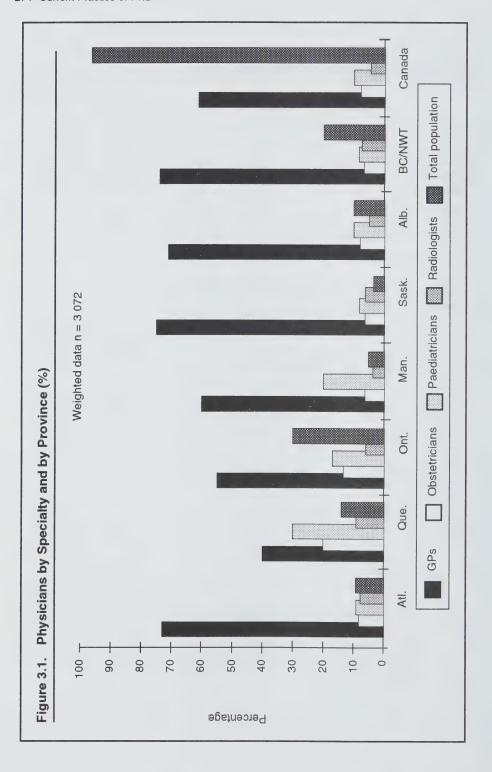
Table 3.1. Study Population by Specialty, Number of Respondents, and Response Rate for Canada as a Whole

	Population	Sample	No. of resp.	Response rate (%)	Weighted no. of resp.
GPs	8 021	1 715	1 045	60.93	1 961.82
Obstetricians	1 528	1 501	773	51.33	372.75
Paediatricians	2 027	1 746	770	44.10	495.02
Radiologists	991	991	484	48.84	242.37
Total	12 567	5 953	3 072	51.60	3 071.96

Table 3.2. Study Population, Number of Respondents, and Response Rate, by Province

	Population	Sample	No. of resp.	Response rate (%)	Weighted no. of resp.
Atlantic	1 244	489	240	49.08	303.05
Quebec	1 943	1 193	738	61.60	474.73
Ontario	4 085	2 461	1`249	50.75	999.15
Manitoba	624	310	151	48.71	152.37
Saskatchewan	588	177	88	49.72	143.81
Alberta British Columbia, Northwest Territories,	1 445	551	253	45.92	353.17
and Yukon	2 638	772	353	51.56	645.19
Total	12 567	5 953	3 072	51.60	3 071.96

of each province. Quebec represented only 16% of the sample, while Quebeckers constitute 25% of the Canadian population. The figures were reversed for the other provinces: Ontario (MDs: 33%; pop.: 38%), British Columbia (MDs: 21%; pop.: 12%), Alberta (MDs: 10%; pop.: 9%), Manitoba (MDs: 5%; pop. 4%), Saskatchewan (MDs: 5%; pop.: 3%), Atlantic provinces (MDs: 10%; pop.: 8%).



Sociocultural Characteristics of Physicians

Female physicians made up 24% of the survey population, their male counterparts 76%. In Saskatchewan, the gap was wider: 14% female and 86% male (A 3.4).17

The mean age of physicians was 44 years. Physicians in the Atlantic provinces were younger (42 years), those in Saskatchewan older (47 years)

(A 3.5).

Some 75% of physicians had English as their mother tongue (17% in Quebec) and 14%, French (76% in Quebec). In 12% of cases, the mother

tongue was neither English nor French (8% in Quebec) (A 3.7).

Twenty-nine percent of respondents gave their religious affiliation as Catholic, 10% Anglican, 13% United Church, 16% other Protestant denominations, 7% Jewish, and 6% other. Nineteen percent claimed to have no religious affiliation. In Quebec, 80% of the sample was Catholic (A 3.8).

Those who actively practised their religion represented 44% of the sample; 30% practised only occasionally, and 26% not at all. Saskatchewan had the largest percentage of physicians practising their religion, Quebec and British Columbia the lowest (33% and 32% respectively) (A 3.9). The majority of practising members were Protestant.

Determining ethnic origin posed various problems of interpretation since designation was entirely up to the respondents. Some 52% of physicians were of "British" origin (including English Canadians), and 16% were "French" (including French Canadians). 18 Physicians of Western European origin other than French and British represented 13% of the sample, Eastern European 9%, Asian 7%, while 3% declared themselves Jewish. In Quebec, 81% of the sample were of French origin (A 3.10).

Professional Characteristics of Physicians

A word about the make-up of the sample with regard to medical specialty will begin this section. Some experts pointed out to us that a number of GPs in English Canada were foreign specialists who had not recertified as specialists since arriving in this country. Upon verification, this does not appear to be the case. Few GPs had studied abroad (14% at most). This is less than the number of specialists who have trained abroad (about 25%). Among GPs who studied outside Canada, only 7.5% had a profile similar to that of the specialists (e.g., number of pregnant women under their care or number of deliveries, ultrasound scans, amniocenteses performed), and represented barely 1% of the GPs in the sample. We also found that foreign-trained GPs whose practice profile was comparable to that of the specialists had attitudes (abortion, directiveness, perception of anomalies) very similar to those of other GPs and unlike those of specialists.

Some 73% of physicians practised in urban areas, 27% in rural settings. In Quebec, there was a higher concentration of physicians in the cities (83%), while in Saskatchewan (36%) and Manitoba (34%) they were

proportionately more numerous in the country (A 3.12).

A large majority of physicians (75%) considered that their clientele was middle-class, 18% that it was underprivileged, and 2% that it was well-todo. More physicians in Saskatchewan (31%) and in the Atlantic provinces (32%) identified their clientele as underprivileged, while in Quebec and in British Columbia the figure was only 11%.

Some 68% of respondents said they were less than 100 km from a genetics centre, 17% between 100 and 250 km, and 15% more than 250 km away. There was a higher percentage of physicians far removed from genetics centres in the Atlantic provinces and British Columbia, the Northwest Territories, and the Yukon (25%) (A 3.15).

Continuing education (58%) and medical journals (43%) were the most commonly cited sources of information on PND, followed by conferences (32%), information passed on by colleagues (31%), and scientific

publications (29%) (A 3.16).19

Respondents were virtually equally divided in styling themselves as "more conservative than liberal" (35%), "more liberal than conservative" (28%), or "equally conservative and liberal" (38%). A higher percentage of physicians in Alberta (46%) saw themselves as conservative than in other provinces (A 3.21).

More than a third of the physicians saw themselves as very directive in their advice to patients. In Saskatchewan, 55% saw themselves in this way (A 3.17).

In general, 77% of physicians said they discussed their opinions with patients, and 84% that they sought the advice of colleagues (A 3.19 and A 3.20). One-third of respondents (30%) said they would readily adopt new technologies; two-thirds would not (A 3.18).

Physicians' Experience with PND

The average number of pregnancies monitored in a year was 44 for GPs and 256 for obstetricians, which also roughly coincided with the average number of deliveries per year (A 3.22 and A 3.23). Quebec GPs monitored more pregnancies and performed more deliveries than their counterparts in other provinces. On average, they attended 86 pregnancies and performed 73 deliveries a year. Paediatricians cared for an average of 246 newborns a year, GPs 40 (A 3.25).

In all, 76% of obstetrical ultrasound scans were carried out by radiologists, 18% by obstetricians, and the remaining 6% by other specialists (A 3.30). Radiologists in the study said they averaged 969 obstetrical ultrasounds a year, obstetricians 402 (A 3.24).

General practitioners ordered an average of 2 amniocenteses, 2 CVSs, and 11 alpha-fetoprotein tests per year, while obstetricians ordered 18 amniocenteses, 7 CVSs, and 72 blood tests. Blood tests were more frequently ordered in Ontario (average, 109 a year) and Manitoba (average,

307 a year): CVS was more frequent in Manitoba, Alberta, and British

Columbia (average, 10 a year) (A 3.26, A 3.27, A 3.28).

Both GPs and obstetricians estimated that about 90% of their pregnant patients had had at least one ultrasound scan. In Manitoba, this was true of only 62% of obstetricians' clientele and 69% of that of GPs (A 3.29). Alberta GPs also had a lower than average percentage: they reported that only 79% of their clientele underwent ultrasound scans.

Summary

In conclusion, although Quebec had a different profile, the typical respondent was a male English-speaking GP, aged 44, whose religion, which he practised moderately, was either Catholic or Protestant. He worked in an urban area, served the middle class, and was close to a genetics centre. GPs cared for about 40 pregnant women a year (twice that figure for their Quebec colleagues), compared to 250 for obstetricians.

Chapter 4. Provincial Differences/Similarities Regarding PND

In this chapter, we present the main areas on which Canadian physicians²¹ agree with regard to the three lines of inquiry covered in this study (use of procedures, perception of anomalies, and social choices), and the main differences between provinces (since the province of practice has proved to be the greatest differentiating factor among Canadian physicians). As mentioned above, detailed tables are contained in Appendix 3, but are referenced throughout the text.

Use of Procedures

Ultrasound

Number

Obstetrical ultrasound scanning has grown exponentially since its introduction and, as various authors have noted, the procedure spread before it was even evaluated (Jacob 1986; Anderson and Allison 1990). Enthusiasm preceded any evidence of its effectiveness or safety. Although it is now a key component of prenatal care, there do not appear to be any explicit empirical standards that would warrant its routine use. Opinions differ on the number of ultrasound scans that should ideally be performed during pregnancy, and indeed on whether there is any valid clinical reason for doing them at all. Consensus conferences, it should be noted, have produced a variety of opinions. In France (Tournaire et al. 1987), the consensus reached was two ultrasound scans per pregnancy. The U.S. National Institutes of Health, on the other hand, indicated in 1984 that there is no evidence justifying a firm and final opinion on this point.

Survey findings show a broad discrepancy between the opinions of physicians from different provinces on the number of ultrasound scans that ideally should be performed during a normal pregnancy (see Figure 4.1). There is a weak consensus among Canadian physicians overall (63%) that at least one ultrasound should be performed, while 20% believe it inappropriate and 16% suggest two ultrasounds. Three provinces figure prominently on the "no ultrasound" scale: Manitoba and Alberta, where 43% and 37% of physicians respectively think no ultrasound scan is necessary, and Quebec, where only 4% of physicians hold that opinion. Quebec physicians are also the most open (38%) to the idea of performing two ultrasound scans during pregnancy (A 4.1).

Reasons

The use of ultrasound scanning can be defended on various grounds, including medical (obstetrical data, detection of malformations) and psychosocial (to reassure the patient, give her a sense of responsibility, learn the sex of the fetus). We wanted to know what importance physicians attached to these various reasons. As may be seen from Figure 4.2, the strongest consensus (86%) related to the procedure's obstetrical usefulness.

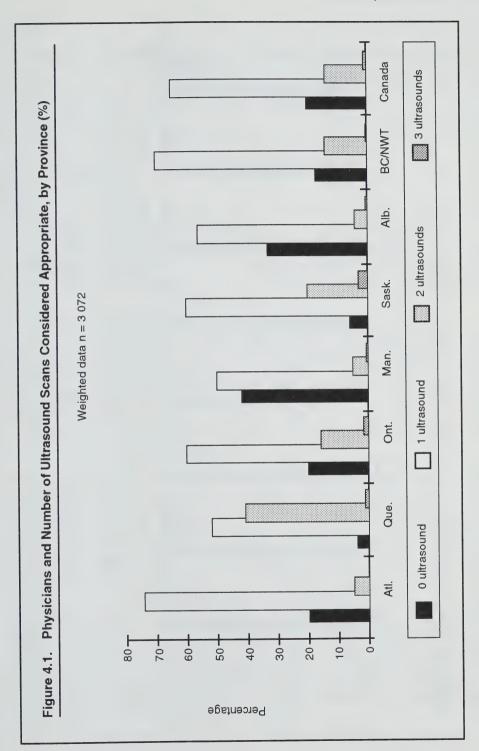
Screening for malformations was considered a valid reason for using ultrasound by 61% of Canadian physicians. Quebec physicians were almost unanimous in their opinion that such use is justified (89%), while in Manitoba and Saskatchewan 40% and 50% of physicians concurred.

One physician in three (28%) thought using ultrasound scans to reassure women was justified (34% in Quebec, 21% in British Columbia). Physicians tended to reject the use of ultrasound scanning for non-medical reasons; using ultrasound to give women a sense of responsibility or learn the sex of the fetus was acceptable only to 12% in the first case, and 7% in the second (A 4.2).

Directiveness of the Physician with Regard to Ultrasound Scanning

In order to determine how important physicians consider the use of ultrasound scanning during pregnancy, we asked them to imagine themselves in a situation where a woman refused the procedure. They were then given three alternatives: accept because the decision is up to the woman, accept because the procedure is unimportant, or reject the woman's decision and refuse to keep her as a patient. As Table 4.1 shows, the great majority of physicians (71%) said that it was up to the woman to decide (78% in Saskatchewan, 76% in British Columbia).

Twenty-seven percent said the procedure was not important (37% in Manitoba and 34% in Alberta). These two provinces, it should be pointed out, are also those where the largest number of physicians believed that ultrasound scanning was not necessary during pregnancy. In Quebec and British Columbia, however, only 20% and 21% of physicians stated the procedure was unimportant.



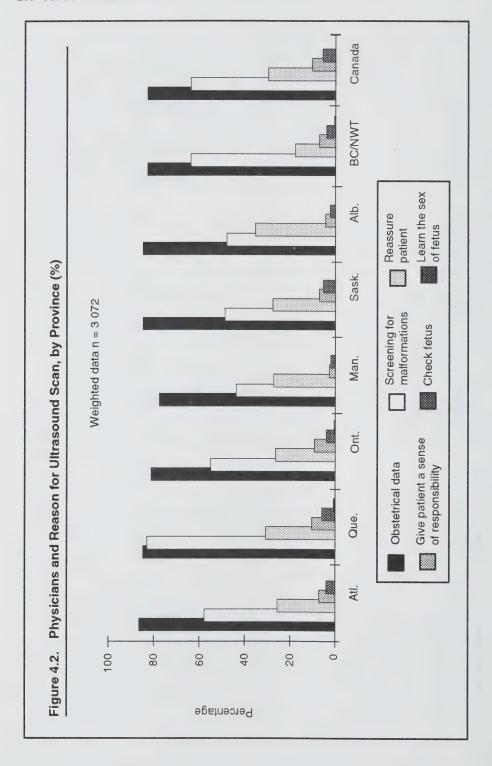


Table 4.1. Physicians' Attitudes if Ultrasound Scan Refused (%) (Q2)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Woman decides Procedure not	69	71	70	63	78	66	76	71
important Would not accept refusal and would suggest referral to	31	20	29	37	22	34	21	27
another physician	1	9	2	-	-	-	4	3

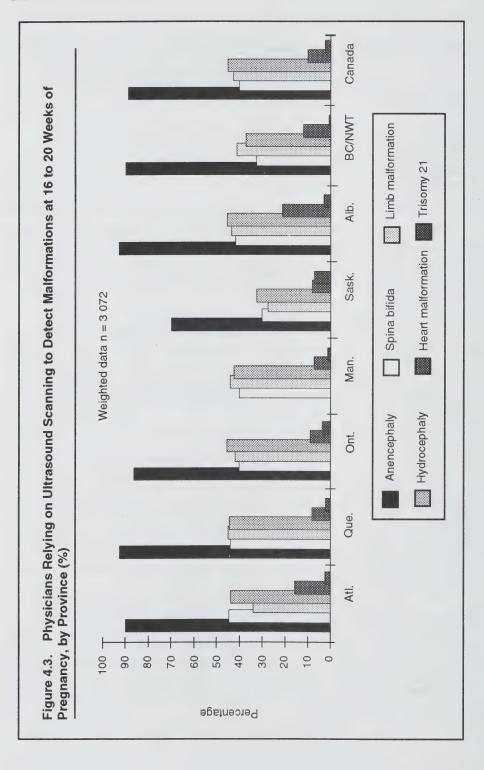
Finally, 3% of physicians (9% in Quebec) would have suggested the woman see another physician. In our Quebec/France study, more than 20% of French physicians in the Picardy/North Pas-de-Calais region said they would have referred the woman to another physician (Renaud et al. 1991) (A 4.3).

Perception of Reliability of Ultrasound Scanning

Given the present state of our knowledge, it is difficult to determine how reliable ultrasound really is in diagnosing anomalies between the sixteenth and the twentieth weeks of pregnancy. The Macquart-Moulin study in France (1989) indicates that ultrasound scans performed between the eighteenth and twenty-second weeks of pregnancy were helpful in diagnosing 73% of anencephalies, 23% of spina bifidas with meningomyelocele, 17% of hydrocephalies, and 37% of limb deformities, but not at all in diagnosing cardiac malformations.

We wanted to know Canadian physicians' attitudes in this regard (see Figure 4.3). If the above-mentioned study reflected Canadian conditions, our medical doctors would overestimate somewhat the reliability of ultrasound scanning in detecting fetal abnormalities. Many physicians, for instance, are totally confident that anencephaly can be detected by ultrasound between the sixteenth and twentieth weeks of pregnancy (88% of Canadian physicians; only 68% in Saskatchewan). About 40% are confident about the diagnosis of hydrocephaly, limb deformity, or spina bifida; 14% about the diagnosis of cardiac malformation.

Only 3% of physicians (12% in Saskatchewan) said they thought ultrasound scanning could be useful in diagnosing trisomy 21. Recent studies emphasize the role of ultrasound in detecting chromosomal anomalies often characterized by deformities. In this case, a thorough ultrasound scan would be a valid approach to determining whether amniocentesis is indicated and would thus improve detection of trisomy 21 (Boué 1989). However, a thorough evaluation has yet to be carried out (Hamerton et al. 1993) (A 4.5).



Amniocentesis and CVS

Preferred Age of Eligibility

There are no rules common to countries with public health insurance systems regarding access to amniocentesis. Some countries, such as France, pay for amniocentesis only if the woman is 38 years of age or over, unless there are medical indications; younger women can nevertheless have their private insurance plans pay for the procedure. Other countries, such as Norway, set a maximum for the number of amniocenteses that can be performed in a given year, leaving it to physicians to decide which cases most warrant it. In yet other countries, such as Great Britain, the age of eligibility for amniocentesis has varied according to hospital budgets. In Canada, the various provincial plans prohibit amniocentesis for women under 35, unless there are medical indications. Women who do not meet these criteria cannot have access to the procedure even if they are prepared to pay for it themselves. In general, laboratories could not meet the additional demand this would put on their services. The same situation exists with regard to CVS, although the age of eligibility is generally higher.

Study findings showed that there was no unanimous agreement on the 35-year limit. A weak consensus (62%) existed for setting eligibility for amniocentesis at 35 years of age (Figure 4.4). Ten percent of physicians would set the limit below 35 years, 23% would set it above that age, and 5% rejected the procedure altogether. Saskatchewan physicians showed the greatest resistance to the threshold of 35 years, with only 44% agreeing with it; they were also the most likely to want the age of eligibility for amniocentesis raised above 35 years (36%) or to reject the procedure entirely (14%) (A 4.6).

Physicians were more divided over CVS (Figure 4.5): 51% of them would set eligibility at 35 years of age, 26% at between 36 and 40, 14% rejected the procedure, and 8% believed it should be made available at any age. Here again, Saskatchewan had the fewest physicians who would accept the norm of 35 years (36%) and the most who would reject the procedure (27%). Quebec physicians matched them closely in this regard, with 23% rejecting the procedure. British Columbia physicians were the most sympathetic to expanding access to amniocentesis and to CVS to women under 35 years of age (15%) (A 4.7).

Access Criteria

Amniocentesis could be used for other reasons besides the mother's age and medical indications: to reduce the expectant mother's anxiety, to learn the sex of the fetus, or simply because the woman is prepared to pay for the procedure. If it *could* be used for those ends, *should* it be?

Another concern is whether amniocentesis should be used in cases where the patient would refuse abortion if an abnormality were diagnosed (Farrant 1985; Fahy and Lippman 1990). Some individuals believe it is a waste of public money to perform the procedure on a woman who would then reject abortion. Others (SOGC 1983) refuse as a matter of principle

to make access to the test conditional on a prior decision to abort. Wertz and Fletcher's (1989b) study on geneticists in 19 countries shows that a majority of them (85% worldwide; 89% in Canada) were in the latter class.

As can be seen from Figure 4.6, only 22% of Canadian physicians — 31% in Quebec — accepted maternal anxiety as a criterion for amniocentesis. This is in contrast to the attitude of the geneticists surveyed by Wertz and Fletcher (1989b) under similar circumstances: 22 73% of geneticists worldwide (70% in Canada) said they would accept such a reason.

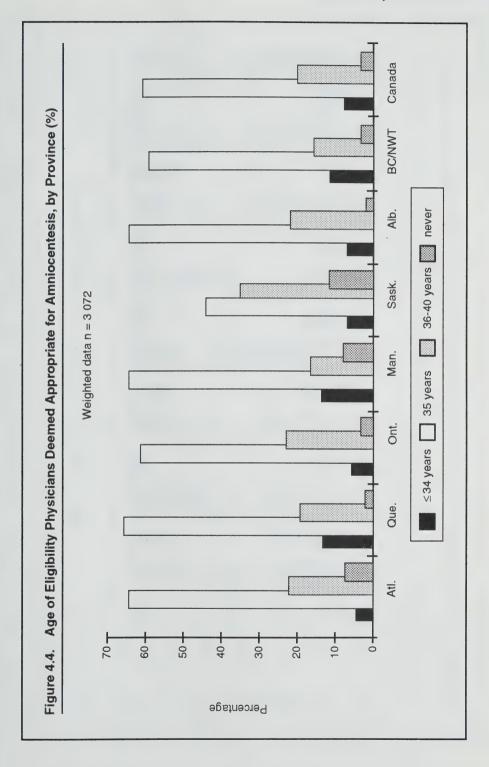
Only 4% of physicians said they would approve access to a procedure like CVS to ascertain the sex of the fetus. This again contrasts with the attitude of geneticists, 42% of whom worldwide (47% in Canada) said they would accept this reason (Wertz and Fletcher 1989b). 23

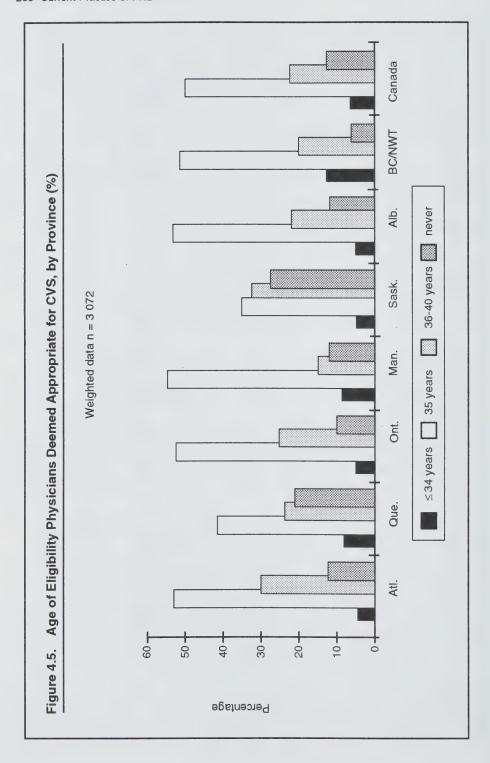
Some 59% of physicians, on the other hand, would agree to make amniocentesis available on the sole criterion of ability to pay. Saskatchewan, the Atlantic region, and Quebec (48%) were least in favour of this position, British Columbia the most (72%). Physicians said, however, that they would not want amniocentesis to be freely available within a publicly financed health care system (75% opposed), just as they would not agree to order an amniocentesis for a 33-year-old woman merely because she requested it.

Medical opinion in Canada, as elsewhere in the world, is very divided on the question of whether amniocentesis should be made available to a woman who would reject abortion in the event of a positive diagnosis. Half the physicians (51%) thought it should not. They consider that the procedure should be made conditional on a prior decision to abort, which is contrary to the opinion of the geneticists in Wertz and Fletcher's (1989b) study. These findings reveal the separation between the principles of sound medical practice and actual practice. Only 36% of physicians (53% in Manitoba) said they thought the procedure should be made available (A 4.8).

Directiveness of the Physician with Regard to Amniocentesis

Faced with a 36-year-old primipara who has misgivings about amniocentesis because of the risk of spontaneous abortion, 45% of physicians would recommend the procedure, 15% would advise against it, 19% would suggest the alternative of a screening ultrasound to identify possible indications for amniocentesis, and 25% said they would propose other measures such as blood tests or would refuse to state an opinion. If the woman was 38 years of age, and hence more at risk for birth of a child with trisomy 21, a slightly higher number of physicians would recommend that she undergo the procedure (63%), but 33% would still recommend ultrasound scanning or some other procedure. In both cases, Quebec physicians, with their higher degree of specialization, would be much more likely than their colleagues in other provinces to recommend the examination (67% if the woman was 36 years of age, 83% if she was 38), and less likely to suggest alternatives (Table 4.2).





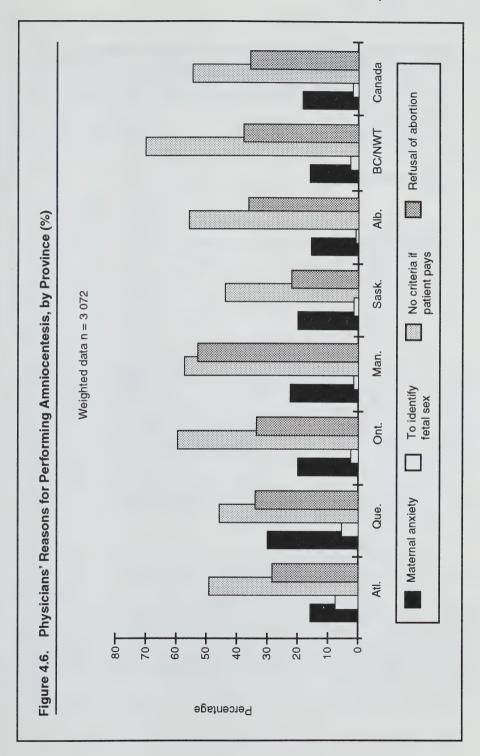


Table 4.2. Physicians' Attitudes Toward a Primipara's Misgivings About Agreeing to Amniocentesis, by Province (%) (Q6-Q7)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
36-year-old woman								
Recommend procedure Do not recommend	36	67	39	42	36	41	49	45
procedure Recommend screening	21	9	13	14	16	14	23	15
ultrasound	19	18	22	15	36	21	10	19
Other	29	7	33	33	14	29	23	25
38-year-old woman								
Recommend procedure Do not recommend	54	83	56	56	53	61	67	63
procedure Recommend screening	11	4	7	6	10	7	10	8
ultrasound	15	9	15	12	27	11	5	12
Other	25	5	27	31	11	25	20	21

All in all, the medical profession has some reservations about access to amniocentesis. Not everyone agrees that the procedure is a necessary part of good pregnancy management. There is a consensus, albeit a weak one, that age 35 should be the threshold, and there is an equally weak consensus that a 38-year-old woman should be encouraged to have the procedure despite her misgivings. Some physicians consider ultrasound scanning and blood tests as alternatives to more invasive procedures. Unlike geneticists, most physicians do not accept anxiety as a valid reason for amniocentesis, and the great majority object to using PND to determine fetal sex. Lastly, they have mixed emotions about making the decision to abort a prerequisite for amniocentesis.

Possibility of Lawsuits and PND

A climate of defensive medicine is developing with the increasing number of lawsuits related to obstetrics and the corresponding rise in insurance premiums. We wanted to know what impact this had had on the physicians surveyed (Table 4.3). For 56% of respondents, the fear of lawsuits would seem to lead to a greater use of PND than would be medically required. The percentage is higher in Saskatchewan (65%) and slightly lower in Quebec (40%) (A 4.10). As will be seen in the next chapter, it is radiologists who fear lawsuits most, followed by GPs, and then by obstetricians.

Table 4.3. Physicians Whose Decision to Use PND Is Influenced by the Possibility of Lawsuits, by Province (%) (Q15 #32)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
For fear of lawsuits we will use PND more often than would be medically indicated	62	40	58	57	65	60	57	56

Predisposition Testing

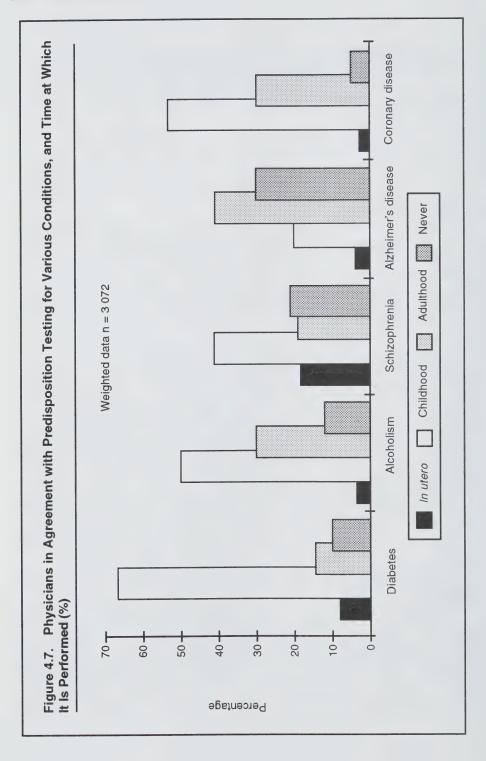
In the future, PND will be used to detect an increasing number of genetic conditions, including predispositions to diseases with multifactorial etiologies. This kind of diagnosis is the subject of constant debate, precisely because it reveals only predispositions that might never materialize. It is difficult to anticipate the implications of these new applications because the uncertainty about their costs and benefits is enormous. There is also serious concern about the discrimination and stigmatization such tests could bring about.

The prospect of offering predisposition testing, at various times in life, for diabetes, alcoholism, schizophrenia, Alzheimer's disease, and coronary heart disease leaves physicians hesitant, as Figure 4.7 shows. Agreement with carrying out such tests *in utero* varied from 3% for coronary heart disease to 17% for schizophrenia. Some 8% of respondents agreed that such tests might be conducted for Alzheimer's disease and diabetes, 4% for alcoholism. Quebec physicians were the most likely to use the tests *in utero* for schizophrenia (27%) and Alzheimer's disease (14%), Manitoba physicians the least likely (4% and 1%).

A fairly large percentage of Canadian physicians, on the other hand, said they would agree to genetic predisposition testing for diabetes (67%), coronary disease (55%), and alcoholism (50%) at birth or in early childhood. The proportion of respondents opposed to such testing at any time varied from 8% for coronary heart disease to 31% for Alzheimer's disease (A 4.11).

Reproductive Practices for Preventing Transmission of Genetic Disorders

In recent years, advances in knowledge and technology have made it possible to introduce various alternative reproductive methods. These methods are designed either to circumvent infertility or to prevent the transmission of genetic disorders. As shown in Table 4.4, the majority of physicians (74%) — slightly fewer in Saskatchewan (64%) and Quebec (66%) — seemed to accept the use of artificial insemination in order to avoid transmitting a dominant disorder. Surrogate motherhood as a way



of countering genetic disease is less well accepted, but is still supported by 40% of medical doctors. These results are consistent with those obtained by Wertz and Fletcher (1989b) for geneticists (A 4.14, A 4.15).

Table 4.4. Physicians in Agreement with Various Reproductive Methods to Circumvent Genetic Disorders (%) (Q15 #16, #22)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Surrogate motherhood	41	35	43	45	33	39	41	40
Artificial insemination	71	66	78	77	64	73	78	74

Acceptability of Sex Selection

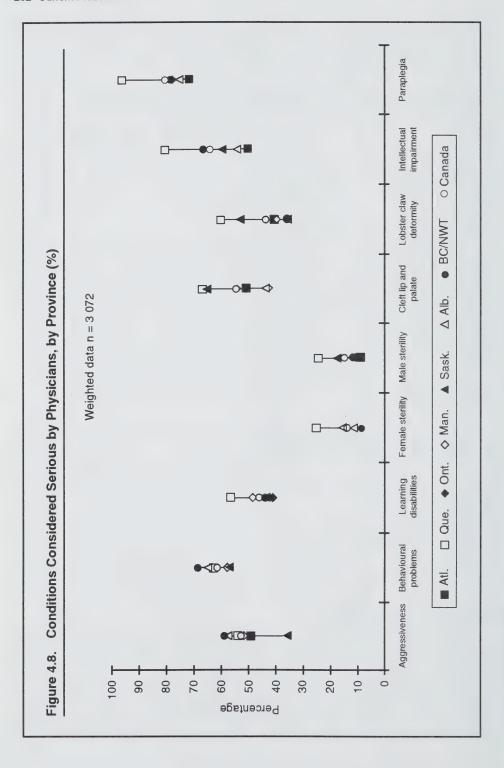
We have seen that physicians are opposed to using PND to determine the sex of the fetus. We wanted to know what they thought of sex selection procedures that produce an embryo of the desired sex and are therefore not associated with abortion (Table 4.5). Only 15% of Canadian physicians said they would find it acceptable to use a chromosome selection method to predetermine the sex of the embryo. Self-prescribed tests to ascertain the sex of the fetus are also clearly unacceptable for the vast majority (86%). Manitoba and Alberta physicians were the most strongly opposed (A 4.16, A 4.17). There is therefore a clear consensus among physicians (based on our definition) against sex selection for non-medical purposes.

Table 4.5. Physicians in Agreement with Various Practices for Selecting/Preselecting the Sex of the Embryo

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Chromosome selection (Q15 #9) Self-prescribed tests to	14	14	17	10	15	9	17	15
ascertain sex of fetus (Q15 #25)	15	12	15	7	10	10	16	14

Perception of Anomalies

Physicians' perceptions of the hardships caused by various anomalies covered a wide range. We asked them how difficult it would be, as a parent, to have a child affected by one of a variety of conditions. Of 15 conditions of varying gravity resulting in either low autonomy (paraplegia, intellectual impairment), potential behavioural problems (e.g., aggressiveness), or sterility (Figure 4.8), paraplegia was perceived as the most difficult (84%). It was followed by behavioural problems (62%),



intellectual impairment (61%), aggressiveness (55%), severe cleft lip and palate (49%), learning disabilities (46%), lobster claw deformity (41%), sex chromosome anomalies (20%-30%), and sterility (15%). Quebec doctors attributed a great deal more gravity to these conditions than did other physicians. The widest difference occurred with regard to intellectual impairment: 84% of Quebec medical doctors considered this condition extremely difficult to live with, as opposed to 50% of physicians in the Atlantic provinces (A 4.18).

We also asked physicians if they would accept the idea of having a child with trisomy 21 themselves; 40% answered in the negative (Table 4.6). Sixty-six percent of Quebec physicians would not accept having a child with trisomy 21, as opposed to only 17% of Saskatchewan physicians (A 4.19).

As for disorders that could someday be candidates for susceptibility testing, schizophrenia topped the list, with 17% of physicians across Canada (27% in Quebec) saying they would recommend *in utero* tests for this condition.

Table 4.6. Physicians Who Would Not Accept Having a Child with Trisomy 21 (%)

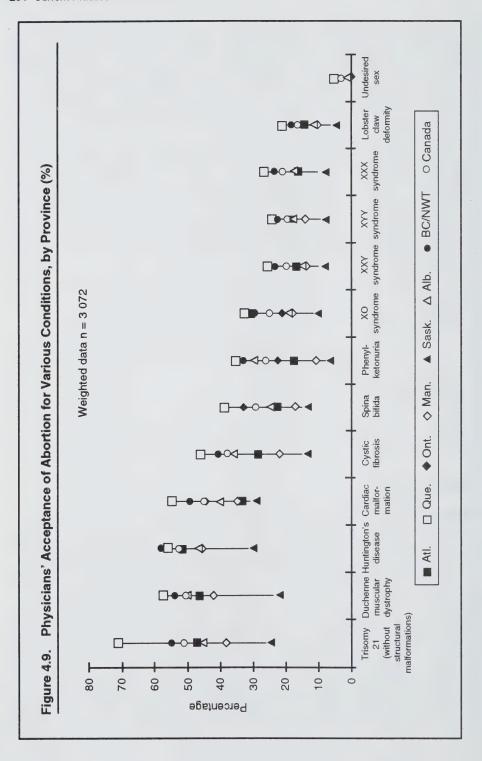
	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
I would not accept the idea of having a child with trisomy 21 (Q15 #30)	32	66	38	25	17	35	41	40

Social Choices

Attitudes Toward Abortion for Various Conditions

As can be seen from Figure 4.9, there is no consensus in favour of selective abortion in Canada, even for trisomy 21, which has been seen by the general public as a legitimate reason for developing amniocentesis.

Fifteen Canadian physicians out of 100 are opposed to abortion following diagnosis of an anomaly, regardless of its nature (see Figure 7.2). This figure coincides with the number of physicians, obtained in other studies on the question (see Table 1.1), who rejected abortion under any circumstances; it is, however, much higher than the more recent figure, obtained, in a similar manner, by our previous surveys (France and Quebec), which indicated that only 5% of medical doctors were unconditionally opposed to abortion (see Table 1.1). The remaining 85% of Canadian physicians were distributed along a normal curve ranging from more or less unsympathetic to sympathetic (see Figure 7.2).



Trisomy 21 without evidence of structural malformations, Duchenne muscular dystrophy, and Huntington's disease are anomalies for which 50% of physicians find abortion acceptable. Generally, since Quebeckers perceive the various disorders as more serious, they tend to be more open to abortion. While aborting a fetus with trisomy 21 without evidence of structural malformations was acceptable to 70% of Quebec physicians, only 25% of Saskatchewan practitioners concurred.

Between 30% and 40% of physicians considered abortion acceptable upon a diagnosis of severe heart malformation, cystic fibrosis, or spina bifida (40%-55% in Quebec). Once again, Saskatchewan doctors were two to three times less likely to find abortion acceptable.

Between 10% and 20% of physicians said they would be agreeable to aborting a fetus with phenylketonuria, a sex chromosome anomaly, or lobster claw deformity (20%-30% in Quebec) (A 4.20).

Respondents were unanimously opposed to aborting a fetus because it was not the desired sex (98% against). If it became possible to diagnose schizophrenia *in utero*, 17% of physicians would favour abortion (27% of Quebec physicians), 7% in the case of Alzheimer's disease (14% in Quebec), and 3% in the case of diabetes (4% in Quebec) (A 4.12).

Nearly half the respondents (47%) said aborting a fetus in the first trimester was more justified than in the second (A 4.22).

One physician in five found it acceptable to encourage a woman with an anencephalic fetus to continue pregnancy so that the fetus's healthy organs could be used for transplants; the percentage reached 30% in Saskatchewan (A 4.25).

Nearly 20% of Canadian physicians said that PND must be condemned if it is performed with the deliberate intention of terminating the pregnancy if an anomaly is discovered (Table 4.7); 70% disagreed. Thirty-three percent of Saskatchewan physicians were prepared to reject PND in such cases, but only 13% of their Quebec colleagues shared their opinion. There thus appears to be a clear consensus (based on our definition) that PND should not be condemned under such circumstances (A 4.24).

Table 4.7. Physicians Who Consider PND Must Be Condemned if the Intention Is to Abort (%)

One must condemn prenatal diagnosis done with the deliberate intention of aborting if the results reveal the existence of an anomaly (Q15 #10) ATL QUE ONT MAN SASK ALB BC/NWT CAN BC/NWT CAN ALB BC/NWT CAN 20 13 19 21 33 21 15 18

In short, selective abortion is far from being completely accepted in Canada. In the country as a whole, 50% of physicians at most agree with selective abortion for certain anomalies (trisomy 21, Duchenne muscular dystrophy, Huntington's disease). There are, however, marked variations between provinces, with Quebec and Saskatchewan at opposite ends of the spectrum. As expected, agreement with abortion on demand is no greater. As Table 4.8 shows, 50% of physicians agree with the statement: "With respect to abortion, parents have an absolute right to freedom of choice."

Table 4.8. Directiveness of Physicians with Respect to the Decision Whether to Abort, by Province (%)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
A physician must be able to resist some abortion requests when he or she considers an anomaly to be minor (Q15 #2)	62	59	62	73	76	65	61	63
Physicians, not parents, should decide which fetal anomalies warrant pregnancy termination (Q15 #6)	18	25	13	19	15	14	13	16
With respect to abortion, parents have an absolute right to freedom of choice (Q15 #4)	43	58	53	40	35	40	52	50

Directiveness of the Physician with Regard to Abortion

Table 4.8 presents findings on various indicators relating to the directiveness of physicians. Their role with regard to the decision whether to abort is the subject of much debate. Under the freedom principles advanced in bioethics and genetics circles, practitioners should in no way intervene in women's decisions. Their role should be limited to supporting, informing, and educating (Wertz and Fletcher 1989b). Others believe that physicians should oppose abortion when a minor, curable abnormality is involved (Maroteaux 1986), and they reflect on which anomalies make abortion justifiable (Clarke 1991). Still others do not consider it possible to maintain a non-directive approach in genetic counselling if anomalies are to be prevented (ibid.). Table 4.8 clearly illustrates Canadian physicians' diverse opinions on the subject.

As noted, 50% of physicians are in favour of giving parents freedom of choice with respect to abortion. The percentage is slightly higher in Quebec (58%) and lower in Saskatchewan (35%). However, even though many

practitioners recognize freedom of choice as an absolute right, a somewhat higher proportion of them believe that "a physician must be able to resist some abortion requests" (63% overall, 76% in Saskatchewan, and 59% in Quebec). On the other hand, when faced with an explicitly directive statement, only 16% of Canadian physicians would go so far as to say that they, and not the parents, should decide which fetal anomalies warrant pregnancy termination (25% in Quebec) (A 4.26, A 4.27, A 4.28).

One respondent in three agreed that abortion must be discussed if the patient is an alcoholic. In British Columbia, the percentage was as high as 40% (A 4.29).

Disclosure of Information

As an ever larger number of disorders of all types are being diagnosed at increasingly early stages, are physicians duty-bound to disclose all the information at their disposal? Although the right to information is a recognized principle, there are particular situations (e.g., mosaicism, sex chromosome anomalies) in which physicians question whether it is appropriate to disclose information that might cause needless worry. Some physicians believe that all information is valuable and must be fully disclosed to the parents. Some physicians also question the relevance of revealing the sex of the baby. In some countries, such as Germany and Denmark, medical associations have gone so far as to issue guidelines suggesting that the sex of the fetus not be revealed before the fourteenth week of pregnancy (Ware 1987; Nippert 1991).

In the case of uncertain diagnoses of XYY, XXY, and XXX syndromes, there is a strong consensus (95%+) in favour of disclosing the information to the carrier parents, even though the physician is not legally bound to do so. On the other hand, 40% of physicians said they sometimes feel obliged to disclose information even though they would prefer not to; this applied to 50% of Saskatchewan physicians (A 4.30, A 4.31).

More than a third of respondents (37%) agreed that, if PND is performed early, as is the case with CVS, the sex of the fetus should not be revealed, except for medical reasons (Table 4.9). Nearly half the respondents in Manitoba (48%) and Saskatchewan (49%) would take this position, compared to only 26% in Quebec (A 4.33).

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Parents having chorionic villus sampling should not be given information on fetal sex unless it is	4							
medically relevant (Q15 #33)	41	26	35	48	49	42	36	37

Perception of the Harmful Effects of PND

As we saw in Chapter 1, PND raises numerous concerns regarding both the intolerance of imperfection that its further development could engender and the discriminatory use to which it could be put. We were able to gauge physicians' attitudes in this regard by means of a series of statements.

Physicians share some of these concerns (Table 4.10). Three respondents out of four (76%) believed there is a danger that the results of predisposition testing will be used for discriminatory purposes. Half of them (49%) considered that the use of PND increases intolerance of even slight anomalies in a fetus or child (A 4.34, A 4.35, A 4.36).

On the other hand, few physicians themselves support eugenics (Table 4.11). Nearly one in three (28%) said that the success of PND is best measured by reductions in the cost of services for the care of children with birth defects. One physician in six (16%) believed that intentionally giving birth to a child with a genetic defect at a time when both PND and abortion are available is socially irresponsible (27% in Quebec). A similar proportion (14%) said they thought it would be justified to enact laws to control the spread of genes causing severe diseases (22% in Quebec). Overall, however, Canadian physicians are opposed to the coercive use of PND — there is a strong consensus (based on our definition) to that effect (75%). Quebec respondents, although opposed for the most part to eugenics, did not show quite the same degree of opposition as their colleagues from other provinces (A 4.37, A 4.38, A 4.39).

Table 4.10. Physicians Who Consider That PND May Be Counterproductive, by Province (%)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
There is a danger that results from tests for genetic predispositions will be used for discriminatory purposes (Q15 #3)	83	67	76	77	71	77	78	76
PND makes disorders out of conditions hitherto considered normal (Q15 #7)	55	47	49	44	57	53	54	51
PND increases intolerance toward anomalies (Q15 #19)	43	59	49	40	60	54	42	49

Table 4.11. Physicians Agreeing with Various Statements Supportive of Eugenics (%)

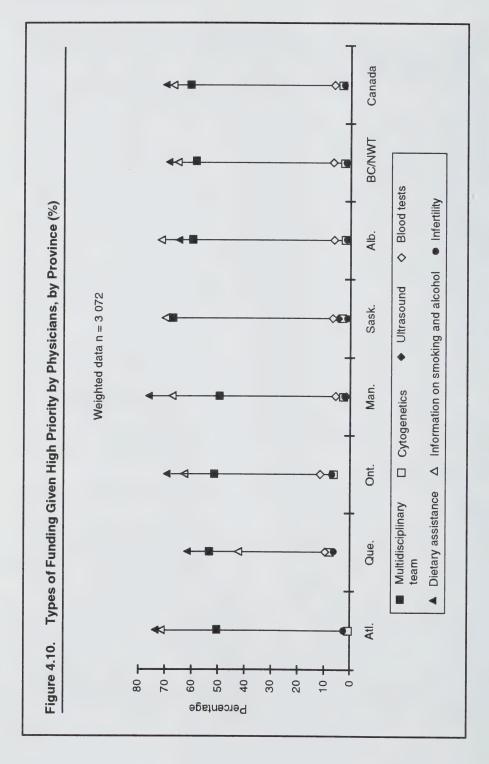
	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Giving birth intentionally to a child with a genetic defect at a time when both PND and abortion are available is socially irresponsible (Q15 #28)	9	27	16	15	9	16	15	16
The success of PND is best measured by reductions in the cost of services for the care of children with birth defects (Q15 #8)	24	32	32	22	17	30	25	28
It would be justified to enact laws to control the spread of genes causing severe diseases (Q15 #31)	9	22	13	12	13	13	11	14

One-half of physicians said they believed that the disabled should be consulted when PND policies are being developed (A 4.40). Twenty percent thought that women rely on PND too much: one physician in three (31%) in Quebec and one in 10 (10%) in British Columbia (A 4.41).

Funding Priorities

The debate surrounding PND is also a debate about funding priorities. How should we allocate the financial resources that society allots to the prevention of anomalies?

Figure 4.10 shows the percentage of respondents in each province who give a high priority to various types of funding (two first choices out of seven). Physicians consider that primary prevention programs (comprehensive dietary assistance and counselling programs to reduce the incidence of low birthweight, funding of multidisciplinary teams in underprivileged areas for pregnant women at risk, information programs on the harmful effects of alcohol and smoking during pregnancy) should have priority over technical screening programs (perfecting cytogenetics, ultrasound training, introducing blood tests for the population as a whole). Preventive programs have the support of 66%, 49%, and 59% of respondents respectively, while only 6% give priority to medical/technical programs. Quebec medical practitioners attach slightly more importance



to developing cytogenetics and obstetrical ultrasound training than their colleagues in the rest of Canada. Treating infertility comes last on the list of priorities (A 4.42).

Only 52% of respondents would favour mass screening for cystic fibrosis if it were possible to identify all carriers. Those from the Atlantic provinces would be the least sympathetic (39%) and those from British Columbia the most sympathetic (59%) (A 4.43). It is also worth noting that 60% of physicians said they thought it important to evaluate the risk of exposure to mutagens and teratogens by means of a prenatal questionnaire (A 4.44).

Forty-six percent of respondents (Table 4.12) were of the opinion that PND cannot be a priority, given the relatively small number of children born with genetic birth defects compared to the much higher percentage who develop serious handicaps due to social or economic factors. Saskatchewan physicians had the highest rate of agreement with this statement (59%), those in Quebec the lowest (37%) (A 4.45).

The great majority (74%) considered they should take the cost of medical services into account in carrying out their work. In the Atlantic provinces, 87% of respondents expressed that opinion; in Quebec the figure was 59% (A 4.46).

Table 4.12. Physicians Considering PND Not a Priority (%)

ATL QUE ONT MAN SASK ALB BC/NWT CAN

PND cannot be considered a priority when only 3% of children are born with birth defects while a much larger proportion born in good health develop serious handicaps caused by social or economic conditions (Q15 #26)

52 37 46 44 59 52 45 46

Summary

Tables 4.13 to 4.16 present a synopsis of the opinions held by those Canadian physicians who responded to the questionnaire. In summary, there is a strong consensus among Canadian physicians (75% or more agree or disagree) on the acceptability of using ultrasound to obtain obstetrical data, the reliability of the procedure to diagnose anencephaly, and its unreliability with regard to trisomy 21. They reject unanimously the idea that amniocentesis should be made available without any criteria being set. They are also strongly opposed to using the procedure for sex selection purposes. The physicians in our sampling clearly believe that all

available information should be disclosed to the parents, even if it is ambiguous.

Sixty percent of respondents agreed on the need for at least one ultrasound scan during a normal pregnancy, on the appropriateness of using the procedure to screen for malformations, on setting the age of 35 as the eligibility threshold for amniocentesis (although this was not so for CVS), and on making amniocentesis available to women who do not meet the normal criteria but are willing to pay for the procedure themselves. Physicians would accept predisposition testing to enable the early treatment of diabetes or preventive counselling for coronary heart disease. A small majority agreed that the fear of lawsuits influences their use of PND.

There is no consensus among physicians across Canada on the acceptability of abortion for a variety of conditions that can be diagnosed *in utero*, or on granting parents freedom of choice on whether to abort. Physicians tend to remain somewhat directive in this respect.

A number of issues are being debated within the medical community: the use of ultrasound scans to reassure women, the reliability of ultrasound scans, the age at which CVS should be available, whether amniocentesis or some other procedure should be suggested as an alternative to women who have misgivings about CVS and whether anxiety is a valid reason for its use, predisposition testing in childhood or in adulthood, funding priorities, the disclosure of fetal sex, and, lastly, the potentially counterproductive effects of PND.

In general, physicians in Manitoba, Alberta, and Saskatchewan are the most conservative about adopting prenatal diagnostic technology. Those in Saskatchewan are the most strongly opposed to the principle of abortion, including the abortion of fetuses with various genetic conditions. At the opposite end of the scale, Quebec physicians are more inclined to use PND procedures and more open to abortion.

	ATL	QUE ONT	MAN	SASK	ALB	BC/NWT	CAN
							%
Ultrasound scanning							
Strongly favour using							
ultrasound to obtain							
obstetrical data							86
Strongly opposed to using							
ultrasound to learn the sex							
of the fetus		**		**			98

	ATL QUE C	ONT MAN SASK AL	LB BC/NWT CAN
Reliability of ultrasound Strongly confident that ultrasound can detect anencephaly		**	88
Very doubtful that ultrasound can detect trisomy 21		**	87
Criteria for access to amniocentesis Strongly opposed to making procedure available without criteria	*	**	75
Strongly opposed to using procedure for sex selection			92
Perceived seriousness of various conditions Paraplegia perceived as very difficult			84
Acceptability of abortion Strongly opposed to aborting a fetus of the undesired sex			98
Information disclosure Strongly opposed to withholding information on sex chromosome anomalies			96
Predisposition testing Strongly agree that testing for predisposition could lead to discrimination	**	**	76

Provinces not indicated form part of the strong consensus.

^{*} Weak consensus (55%-64%).** Moderate consensus (65%-74%).

Table 4.14. Moderate Consensus and Differences Between Provinces

ATL QUE ONT MAN SASK ALB BC/NWT CAN %

Ultrasound scanning Moderately favour accepting refusal of the procedure because the decision is up to the woman * * * * * * 71

Moderately favour accepting refusal of the procedure because the decision is up to the woman				*	**		**	71	
Moderately opposed to using ultrasound as a means of giving women a sense of responsibility		*			*	*	*	65	
Predisposition testing Testing moderately acceptable - in childhood, for diabetes - to enable preventive counselling for alcoholism	*		*	*				67 72	
Procedures to circumvent genetic diseases Artificial insemination moderately acceptable			**	**	*		**	74	
Sex selection Moderately opposed to predetermining sex				**		**		72	
Moderately opposed to allowing self-prescribed tests to determine fetal sex	*	**	*			**		69	
Acceptance of abortion Moderately opposed to aborting a fetus with - lobster claw deformity	**	*		**	**	**			
- a minor malformation					**			74 71	
Moderately opposed to condemning PND if there is a deliberate intention to abort		**					**	72	

Table 4.14. (COTTE O	Table	4.14.	(cont'd)
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ATL QUE ONT MAN SASK ALB BC/NWT CAN % Counterproductive effects of PND (eugenics) Moderately opposed to considering refusal to use PND and abortion as socially irresponsible Moderately opposed to the enactment of laws to control the spread of 74 deleterious genes **Funding priorities** Moderately opposed to physicians not taking into account the cost of medical 74 services

Provinces not indicated form part of the moderate consensus.

Table 4.15.	Weak Consensus and	Differences	Between	Provinces

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN %
Ultrasound scanning Weak consensus in favour of one ultrasound	**						*	63
Low acceptance for using it to detect malformations		**		?	?	? .		61
Weak opposition to making ultrasound subject to written prior agreement								59

Weak consensus (55%-64%).

^{**} Strong consensus (75% and above).

[?] Debatable.

Table 4.15. (cc	nt'd\
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	ATL	QUE	ONT	MAN	SASK AI	LB BC/NWT	CAN %
Reliability of ultrasound Weak consensus that ultrasound is not reliable to detect a heart malformation							57
Criteria for access to amniocentesis Weak agreement with eligibility at age 35	*	*			?		62
Weak agreement with making procedure available without criteria if patient pays	?	?			?	*	59
Weak agreement with recommending procedure to 38-year-old woman who has misgivings	?	**			?	*	62
Weak opposition to giving a 33-year-old woman the choice of having the procedure						*	57
Impact of lawsuits Weak consensus that fear of lawsuits might lead to PND being used more often than medically required		?			*		56
Predisposition testing Weak agreement with		·					
testing for: - early treatment of diabetes - preventive counselling for coronary heart disease		?			**	*	60 59
Perception of gravity Weak consensus that it is difficult to live with:							
- aggressiveness - behavioural problems - intellectual impairment	?	**		?	?	*	55 62 61

Table 4.15. (cont'd)

	ATL	QUE	ONT	MAN	SASK	ALB BC/NWT	CAN %
Weak consensus that it is not difficult to live with: - female sterility - male sterility	*	?				*	59 61
Acceptability of abortion Weak consensus that it is not acceptable to abort a fetus with: - phenylketonuria - Turner's syndrome - Klinefelter's syndrome - XYY syndrome - XXX syndrome	*	???	*	**	** * * * **	*	63 60 63 64 65
Weak agreement that elective abortion is less acceptable than selective abortion							56
Directiveness with regard to abortion Weak agreement that physicians should resist some abortion demands				*	**	*	63
Weak opposition to the physician deciding which anomalies warrant abortion							60
Funding priorities Weak consensus on the importance of evaluating the risk of exposure to mutagens and teratogens during pregnancy						*	60

^{*} Moderate consensus (65%-74%).

Provinces not indicated form part of the weak consensus.

^{**} Strong consensus (75% and more).

[?] Debatable.

	ATI	OUE	ONIT	BAAR	CACK	ALD DOWNER	CAN
	AIL	QUE	ONI	MAN	SASK	ALB BC/NWT	%
Ultrasound scanning Using ultrasound to reassure women							29
Accepting refusal of ultrasound because the procedure is not important							27
Reliability of ultrasound Confident that it can diagnose: - spina bifida - limb malformations - hydrocephaly							39 42 45
Eligibility criteria for amniocentesis and CVS Age of eligibility for CVS should be 35				*		*	51
Recommending to a 36- year-old woman who has misgivings that she should have the test		**					45
Anxiety is not a valid							

Ultrasound scanning Using ultrasound to reassure women								29
Accepting refusal of ultrasound because the procedure is not important								27
Reliability of ultrasound Confident that it can diagnose: - spina bifida - limb malformations								39 42
- hydrocephaly Eligibility criteria for amniocentesis and CVS Age of eligibility for CVS								45
should be 35 Recommending to a 36- year-old woman who has misgivings that she should have the test		**				*		51 45
Anxiety is not a valid criterion of eligibility for amniocentesis	*		*		*	*	*	54
The procedure should not be available if a woman refuses to consider abortion	*							51
Predisposition testing Acceptability of testing for: alcoholism, in childhood								50
schizophrenia, in childhood Alzheimer's disease, in adulthood				*	*		*	44 42
Acceptability of reasons for testing for: - early treatment of								
schizophrenia - Alzheimer's disease				*	*			46 30

Point of predisposition testing if no treatment is available Methods to circumvent genetic disease Acceptability of surrogate motherhood Perception of gravity These conditions are hard to live with: - learning disabilities - hypogonadism - XXX syndrome	*			*	50
genetic disease Acceptability of surrogate motherhood Perception of gravity These conditions are hard to live with: - learning disabilities - hypogonadism - XXX syndrome	*				40
These conditions are hard to live with: - learning disabilities - hypogonadism - XXX syndrome	*				
learning disabilitieshypogonadismXXX syndrome	*				
- hypogonadism - XXX syndrome					46
- XXX syndrome					22
					24
- XXY syndrome					28
- XYY syndrome					22
- cleft lip and palate	*		*		49
- lobster claw deformity - trisomy 21, without	*				41
structural malformations Attitudes to abortion	*				40
Acceptability of abortion for - trisomy 21 without					
structural malformations - Duchenne muscular	**				51
dystrophy	*				51
- Huntington's disease	*				51
- severe heart malformation - cystic fibrosis					43
(unacceptable -) - spina bifida			**(-)		37
(unacceptable -)		*(-)	*(-)	*(-)	29
Abortion in first trimester					
more justifiable than in second					47
Unacceptability of encouraging women with an anencephalic fetus to					
continue their pregnancy so					
the fetus's healthy organs can be used	*				

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	ATL	QUE	ONT	MAN	SASK	ALB BC/NWT	CAN %
Directiveness in relation to abortion Parents' freedom of choice with regard to abortion is an absolute right (do not agree -)		*			*(-)		50
Unacceptability of discussing abortion with alcoholic women					*		42
Disclosure of information Not revealing the sex of the fetus when PND is early							37
Feeling legally bound to disclose information when would rather not							40
Perception of counterproductive effects of PND PND makes disorders out of conditions that would otherwise be considered normal	*				*		51
PND increases intolerance to abnormality		*			*		49
The success of PND lies in the reduction in the cost of caring for sick children							29
PND policies should be developed in consultation				*			53
Women put too much faith in PND							20
Funding priorities PND cannot be considered a priority when the number of birth defects is compared to handicaps due to social							
factors					*		46

AT	L QUE	ONT	MAN	SASK	ALB BC/NWT	CAN %
Acceptability of mass screening for cystic fibrosis					*	52
Weak consensus (55%-64%) Moderate consensus (65%-74) Debatable.						

Chapter 5. Major Differences Between Medical Specialties

The social sciences are surprisingly silent on the psychosocial and sociological profile of the various medical specialties: this was apparently a popular topic in the 1960s (Bucher 1962; Bucher and Strauss 1961), but subsequent generations failed to pursue it. Yet members of different specialties, although they are all physicians, can have very different attitudes, depending on the particular relationship they have with their work, disease, technology, their patients, and their colleagues. Radiologists, for instance, are by definition "technical" people whose contacts with patients are infrequent and indirect. Obstetricians focus on mothers, the course of pregnancies, birth, and experimenting with new methods and procedures. Their main concern is to ensure that the pregnancy has the best possible outcome, and, unlike paediatricians and GPs, they are not responsible for medical follow-up of disabled children (Clarke 1991). GPs and paediatricians are closer to families and their problems. Educating, counselling, and providing information often occupy much of their time (Bartholome 1987). They are also less subject to the imperatives of technological development.

This description is, of necessity, very superficial, but it can nevertheless be assumed that members of the same medical specialty, by virtue of the fact that they share a work environment, often come to develop a common vision of medical technology (distinct from that of other specialties), of the physician-patient relationship, funding priorities, and solutions to health problems. They also acquire a particular body of knowledge, skills, and expertise, and they view PND from the angle of their chosen field of practice.

The few studies on the subject concern the attitude of various specialties toward abortion. It appears that the more familiar physicians

are with the problems experienced by women, the greater is their tolerance of abortion (Bourne 1972). Some specialties are more inclined to protect the interests of women (obstetrician-gynaecologists), while others have the interests of the fetus as their focal point (paediatricians and radiologists) (Fellous 1991). GPs, who are less research-oriented and more involved in clinical work, appear less sympathetic than other physicians to abortion, amniocentesis, and genetic counselling (Carlos and Cloutier 1976; Weitz 1979; Lalardrie 1991).

Use of Procedures

Ultrasound Scanning

Number

The number of ultrasound scans that physicians considered appropriate in the course of a normal pregnancy varied considerably depending on their specialty. While 23% of GPs and 18% of paediatricians said they believed no ultrasound scanning is required during a normal pregnancy, only 12% of obstetricians and 8% of radiologists shared that opinion. Nine percent of GPs believed that two ultrasound scans are needed, compared with 30% of paediatricians and radiologists (A 5.1). These differences are illustrated in Figure 5.1.

Reasons

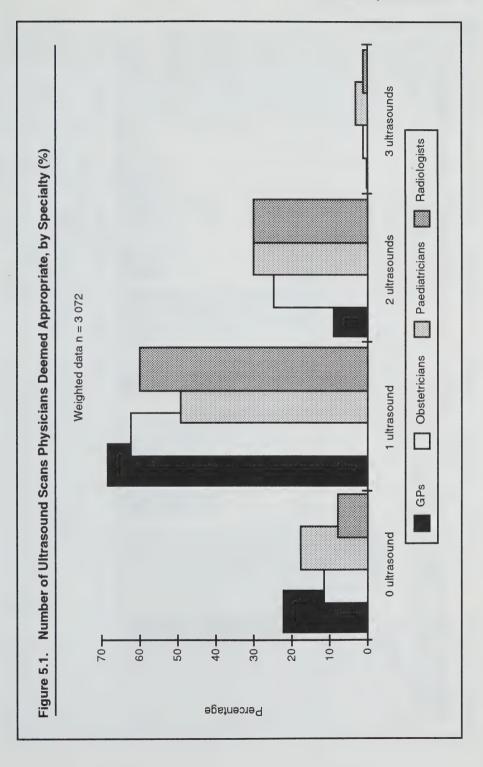
Physicians in the four specialties generally had the same perception of the acceptability of various reasons for using ultrasound technology. However, while there was a positive consensus among obstetricians (75%), paediatricians (70%), and radiologists (80%) on the use of ultrasound scanning to screen for malformations, only 54% of GPs viewed this as a valid reason (A 5.2).

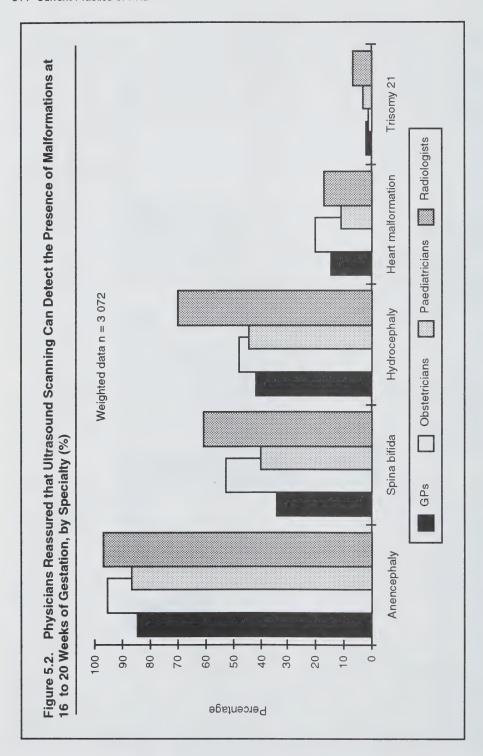
Directiveness of Physicians with Respect to Ultrasound Scanning

The attitude of obstetricians and radiologists toward a patient who refuses ultrasound differed markedly from that of GPs, as indicated by the various reasons for finding a refusal acceptable. Whereas obstetricians (79%) and radiologists (83%) were more inclined than GPs (68%) to accept a refusal because the decision is up to the woman, GPs (30%) were more inclined than obstetricians (18%) and radiologists (11%) to accept a refusal because they are not convinced the procedure is essential (A 5.3, A 5.4).

Perception of Reliability of Ultrasound Scanning

The perception that physicians have of the reliability of ultrasound in detecting various anomalies at 16 to 20 weeks also varies markedly with their specialty, as can be seen from Figure 5.2. In general, radiologists and obstetricians were more reassured by the results of an ultrasound scan than were GPs and paediatricians. For anencephaly, almost all radiologists (98%) and obstetricians (94%) said they were totally reassured, but only 86% of GPs shared their conviction. In addition, there was a consensus only among radiologists on the reliability of ultrasound scanning in





detecting hydrocephaly and spina bifida. Seventy percent said they were reassured in the case of hydrocephaly and 60% in the case of spina bifida, while the percentage is only 45% and 40% for Canadian physicians overall. The percentage of physicians who said they would be reassured with regard to heart malformations was greater for obstetricians (20%) than for physicians as a whole (14%). Finally, radiologists were twice as likely (7.5%) to be reassured about trisomy 21 results than other physicians (3.2%) (A 5.5).

Amniocentesis and CVS

Desired Age for Access

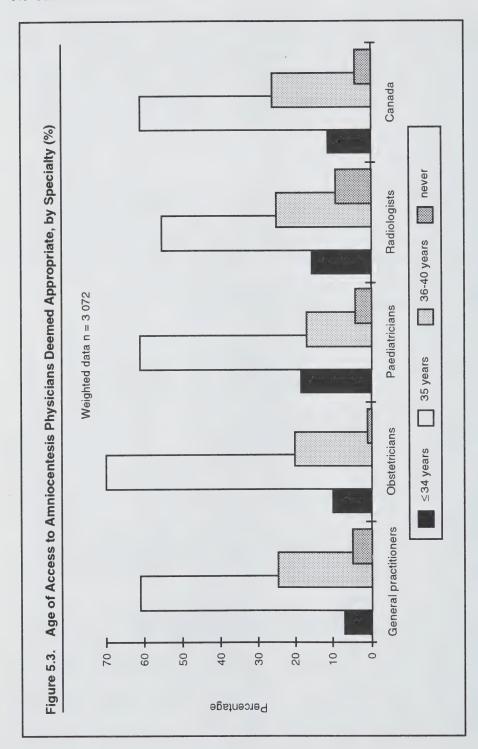
As Figure 5.3 shows, a solid majority of Canadian physicians (62%) agreed that women with no family history of problems should be eligible for amniocentesis at 35 years of age. Nearly twice as many paediatricians (18%) and radiologists (16%) as GPs (7%) and obstetricians (9%) said they would recommend the procedure before that age. Canadian medical practitioners are slightly more divided in their opinions regarding the eligibility threshold for CVS (51% of them suggested age 35), but the differences between specialties remain minor (A 5.6, A 5.7).

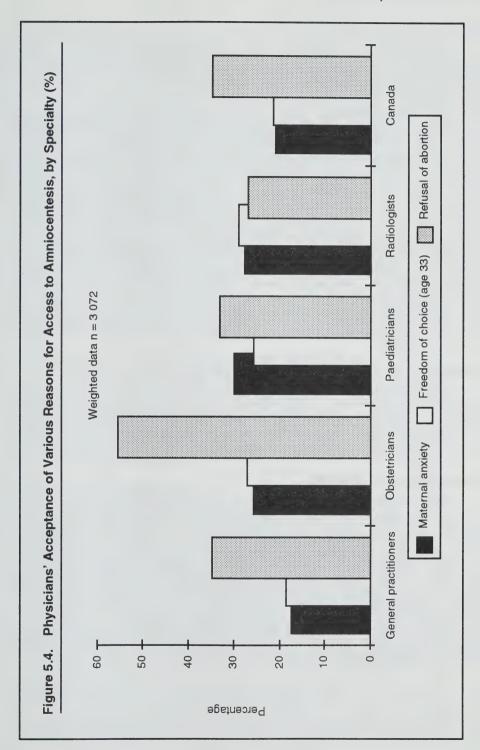
Eligibility Criteria

Most non-medical reasons for using amniocentesis are judged unacceptable by the majority of Canadian physicians, as shown in Figure 5.4. On the other hand, acceptability of such reasons varies greatly by specialty. Compared to physicians in other specialties, and radiologists in particular. GPs are less in favour of expanding the eligibility criteria for the procedure. Thus. 29% of radiologists and 30% of obstetricians, but only 18% of GPs, believe that relieving the anxiety of a 33-year-old woman with no specific history of problems is an acceptable reason for amniocentesis. While 29% of radiologists and 28% of obstetricians recognize freedom of choice as valid grounds for the procedure, only 19% of GPs share that opinion. Radiologists (19%) and paediatricians (24%) are more inclined than obstetricians (14%) and GPs (11%) to believe that amniocentesis should be available to all women, regardless of their age or status. Lastly, a majority of obstetricians (55%), compared to 34% of GPs and 27% of radiologists, said they would find it acceptable to perform amniocentesis on a woman who said she did not want to abort if the fetus proved abnormal (A 5.8).

Physicians' Directiveness with Regard to Amniocentesis

If faced with a woman who had misgivings about amniocentesis because she feared spontaneous abortion, many more radiologists than other physicians would recommend that she have an ultrasound scan: 35% of radiologists as opposed to 18% of all respondents when the woman is 36 years old, and 24% of radiologists as compared to 11% of Canadian physicians when the woman is 38. GPs are more inclined than their colleagues not to recommend the test: 19% versus 7% if the woman is 36, and 9% versus 4% if she is 38 (A 5.9).





Impact of Lawsuits on Use of PND

The proportion of physicians who answered that fear of lawsuits made them use PND more often than medically indicated was greater among radiologists (63%) and GPs (59%) than for obstetricians (49%) and paediatricians (45%) (A 5.10).

Predisposition Testing

Overall, however, a great majority of physicians are opposed to testing for genetic predisposition. If testing became available for diabetes, alcoholism, schizophrenia, Alzheimer's disease, and coronary heart disease, GPs (15%) would be clearly less in favour of using it prenatally than obstetricians and, to a lesser extent, radiologists. Paediatricians adopted a middle-of-the-road position. As regards schizophrenia, GPs (15%) were less favourable to prenatal testing for predisposition than other physicians (23%) and, more particularly, obstetricians (27%). The same was true for Alzheimer's disease, with 6.4% of GPs in favour of testing, as opposed to 12% of other physicians and 13% of radiologists. For diabetes, 5.5% of GPs would be in favour of prenatal testing versus 14% of obstetricians and 12% of radiologists (A 5.11). It is clear (A 5.12) that these tests are accepted mainly with a view to possible early treatment or preventive counselling and that GPs are the most opposed to their being used with the intention of preventing births.

Acceptability of Sex Selection Procedures

In general, a large majority of physicians (7 out of 10) said they found unacceptable procedures that make it possible to select the sex of the embryo. Proportionately more obstetricians (22%) than Canadian physicians in general (15%) are favourable to the idea of predetermining the sex of the embryo through chromosomal selection (A 5.13, A 5.14).

Perception of Anomalies

Members in all four specialties generally have very similar perceptions of the seriousness of the many problems and syndromes presented in our survey. Paediatricians see behavioural problems as less serious than do other physicians, with 52% of them, as opposed to 62% of all physicians, viewing those problems as very serious. Furthermore, while radiologists agree that a severe case of cleft lip and palate is very serious (67%), the opinion of other physicians regarding this problem is much more divided (about 46% see the problem as very serious) (A 5.15).

As for accepting the idea of having a child with trisomy 21 themselves, obstetricians (59%) and, to a lesser extent, radiologists (52%) are more unaccepting than GPs (35%) (A 5.16).

Social Choices

Attitude Toward Abortion for Various Conditions

Physicians' attitudes varied regarding abortion for the fetal anomalies being studied. Except for Turner's syndrome, GPs are less receptive to abortion than other physicians. Obstetricians, on the other hand, are more open to the procedure than their colleagues, regardless of the anomaly involved. Radiologists and paediatricians concur with obstetricians on five and six conditions respectively of the 12 covered (A 5.17). GPs are the most strongly opposed of all physicians to genetic predisposition testing for the purpose of preventing births. Figure 5.5 shows the percentage of physicians accepting of abortion, by anomaly and by specialty. It is evident that, of the four specialties, GPs are consistently more conservative and obstetricians are consistently more open to abortion.

In general, the greater the familiarity with a problem, the greater its seriousness for a specialty and the greater the openness to abortion in that particular case. The vast majority of obstetricians thus accepted pregnancy termination in the case of trisomy 21 (70% versus 51% for physicians in general). Similarly, radiologists accepted abortion more readily for severe heart malformations (62% versus 43% overall), and paediatricians for cystic fibrosis (50% versus 37% overall).

Physicians' Directiveness with Respect to Abortion

As regards directiveness, Table 5.1 shows that obstetricians are the most inclined to give parents total freedom of choice on terminating a pregnancy; 61% favour such rights for parents, as opposed to 50% of Canadian physicians in general. A larger proportion of paediatricians (21%) than Canadian physicians in general (16%) believe that physicians, not parents, should decide which anomalies warrant pregnancy termination (A 5.22, A 5.23).

Disclosure of Information

Asked whether physicians should tell parents when a fetus has a sex chromosome aberration, the vast majority of respondents (96% on average) answered in the affirmative. It should be noted, however, that proportionately more radiologists than other physicians answered that such information should be withheld: 9.8% of radiologists versus 5.6% of all Canadian physicians for the XYY syndrome, 5% as opposed to 2% for the XXY syndrome, and 8% versus 4% for the XXX syndrome. More radiologists also believed that information should be withheld if they considered the anomaly to be minor (11% versus 5% for all physicians) or when it involved early diagnosis of the embryo's sex, except for medical reasons (44.4% versus 36.5%) (A 5.24, A 5.25, A 5.26, A 5.27).

Perception of Counterproductive Effects of PND

On the whole, specialties have similar attitudes regarding the potentially counterproductive effects of PND (discrimination, intolerance, turning normal conditions into pathological ones). However, a larger

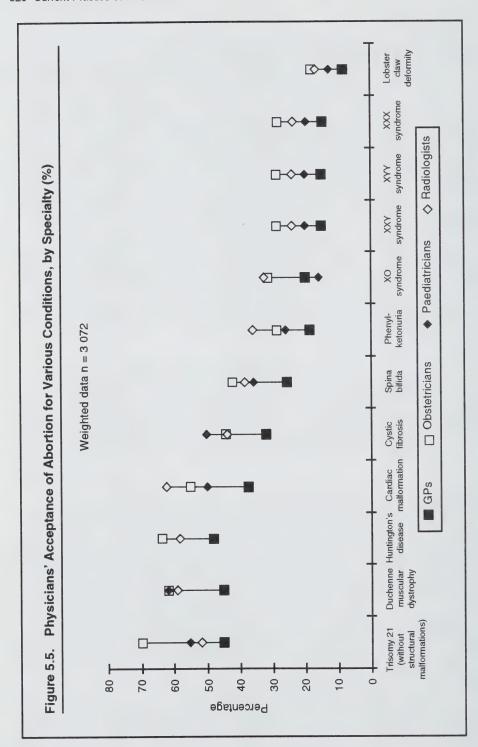


Table 5.1. Physicians' Directiveness with Regard to Abortion, by Specialty (%)

	GPs	Obstetricians	Paediatricians	Radiologists	CAN
A physician must be able to resist some abortion requests when he or she considers an anomaly to be minor (Q15 #2)	60	57	75	67	63
Physicians, not parents, should decide which fetal anomalies warrant pregnancy termination (Q15 #6)	14	15	21	15	16
With respect to abortion, parents have an absolute right to freedom of choice (Q15 #4)	48	61	49	51	50

proportion of GPs (56%) than members of other specialties (43%) agreed that developments in PND will lead to previously normal conditions being seen as disorders (A 5.28).

The majority of physicians were clearly not supportive of the coercive or discriminatory use of PND, judging by their answers to the three questions on this issue. The GPs' position is even more clear-cut than that of their colleagues. Only 12% of GPs, compared to 23% of obstetricians, 20% of paediatricians, and 30% of radiologists, consider that intentionally giving birth to a child with a genetic defect at a time when PND and abortion are available is socially irresponsible. Asked whether laws should be enacted to control the spread of genes causing severe diseases, 11% of GPs and 13% of paediatricians answered in the affirmative, compared to 21% of obstetricians and radiologists (A 5.29, A 5.30).

Funding Priorities

Allocation of the health care budget did not differ fundamentally by specialty. However, obstetricians said they would attach less importance to evaluating the risk of exposure to mutagens and teratogens than their Canadian colleagues; 46% of obstetricians attach a great importance to it, compared to 60% of all Canadian physicians. More GPs (52%) than specialists (36%) agreed that PND should not be considered a priority, given the small percentage of children with birth defects and the far larger

percentage of children who are born healthy but develop serious handicaps as a result of social factors (A 5.31, A 5.32).

Summary

Table 5.2 gives an overview of the main differences between members of different specialties. There is a consistent divergence between GPs and specialists, most notably obstetricians and radiologists; however, the differences we observed were mostly small (10%-20% on average).

The variations observed were nevertheless statistically significant, consistent and systematic. Compared to other physicians, GPs were less favourable to the use of PND procedures (ultrasound scanning and amniocentesis), less favourable to expanding their scope (eligibility criteria for amniocentesis, use of prenatal testing for predisposition, allocation of health care funds to PND), and more wary of their reliability and effects on society. GPs were also less open than their colleagues to pregnancy termination for various fetal anomalies and less supportive of the coercive or discriminatory use of PND. At the opposite end of the spectrum, obstetricians — and radiologists even more so — placed greater trust in PND technology and were consistently more in favour of its use and development. These physicians - more particularly obstetricians, and paediatricians to a lesser extent — were more inclined to justify pregnancy termination for various fetal anomalies. Obstetricians had a more sympathetic attitude toward sex selection and a less favourable opinion about evaluating the risk of exposure to mutagens and teratogens. Radiologists, on the other hand, had greater directiveness and a protechnology stance when faced with ambivalent patients, and a stronger opposition to the disclosure of information. Paediatricians differed from other specialties mainly because of their middle-of-the-road position, sometimes leaning toward GPs and sometimes toward obstetricians and radiologists.

To sum up, the various medical specialties have differing attitudes to PND. While the differences are not large, they are nonetheless consistent. GPs are more socially oriented in their choices, less inclined to use PND procedures, and more conservative about their expansion and about abortion. Obstetricians and radiologists, on the other hand, are more in favour of the procedures. We can, therefore, observe that the various medical specialties exhibit attitudes resulting from a slightly different view of the world.

Table 5.2. Significant Differences, by Medical Specialty

	GPs	Paediat.	Obst.	Radio.	Tot. pop.
Ultrasound scanning					
No ultrasound should be done	+	+	_	_	20
Two ultrasounds should be done	_	+	+	+	16
Use ultrasound to screen for					
malformations	-	+	+	+	61
Agree to refusal of ultrasound because					
decision is the woman's	-	+	+	+	71
Agree to refusal of ultrasound because					
scan is not important	+	-	-	-	27
Reliability of ultrasound scanning					
Confidence in ultrasound to detect:					
anencephaly	_	_	+	+	88
hydrocephaly	_	+	·	+	45
spina bifida	_			+	39
trisomy 21				+	3
•					
ligibility criteria for amniocentesis					
Expand access to women aged 35 years					
and under	_	+	-	+	10
Anxiety is a valid eligibility criterion for					00
amniocentesis	_	-	+	+	22
Agree to 33-year-old woman's freedom of choice to have procedure					22
Make amniocentesis available without			+	+	22
viake ammocentesis available without criteria					14
Make test available even if woman	-	+	_	+	14
ejects abortion			+		36
ejects abortion	_			_	30
Directiveness with regard to					
ımniocentesis					
Recommend going through with					
procedure (36-year-old woman)				+	45
Recommend going through with					
procedure (38-year-old woman)				+	63
Recommend not having procedure					
age 36 and 38)	+				11
mpact of lawsuits					
More frequent use of PND because of					
ear of lawsuits	+	_	***	+	56
				•	

	GPs	Paediat.	Obst.	Radio.	Tot. pop.
Predisposition testing					
In favour of prenatal genetic					
predisposition testing to detect:					
- schizophrenia	-	+	+	+	17
- Alzheimer's disease	_	+	+	+	9
- diabetes	-		+	+	8
In favour of testing to prevent births					
(schizophrenia)	-	+	+	+	17
Sex selection procedures					
Predetermining fetal sex by					
chromosomal selection			+		15
Perception of gravity of various					
conditions					
These conditions are seen as serious or	•				
difficult:					60
- behaviour problems	+	_		,	62 49
- severe cleft lip and palate		_	_	+	49
Acceptability of abortion					
Abortion acceptable in case of:					
- trisomy 21	-		+		51
- Duchenne muscular dystrophy	_	+	+		51
- Huntington's disease	_	+	+	+	51
- severe heart malformations	-	+	+	+	43
- cystic fibrosis	-	+	+		37
- spina bifida	-	+	+	+	29
- phenylketonuria	_		+	+	22
- Turner's syndrome		-	+	+	21
- Klinefelter's syndrome	_		+		17
- XYY syndrome	_		+		16
- XXX syndrome	-		+		16
- lobster claw deformity	_		+	+	10
Directiveness in regard to abortion					
Freedom of choice of parents regarding					
abortion is absolute	_		+		50
Physicians should decide which					
anomalies warrant abortion		+			16

Table	5.2.	(cont'd)
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	GPs	Paediat.	Obst.	Radio.	Tot. pop. (%)
Information disclosure Favour revealing that fetus has a sex chromosome anomaly					96
Favour disclosing information even if anomaly is minor				_	88
Perception of the counterproductive effects of PND PND makes disorders out of conditions that would be considered normal Giving birth to a child with a genetic defect at a time when prenatal diagnosis and abortion are available is socially irresponsible It is justified to enact laws to control the spread of genes causing severe diseases	+	+	-++	+	51 16 14
Funding priorities Favour evaluating the risk of exposure to mutagens and teratogens PND is not a priority given the percentage of birth defects compared to that of acquired problems due to social factors	+		-		15 46

Only statistically significant differences are indicated.

Tot. pop. = percentage of physicians in favour for Canada as a whole.

= above Canadian percentage.

= below Canadian percentage.

Chapter 6. Major Differences, by Religion and Religious **Practice**

Many other factors in addition to province of practice and specialty can help us understand physicians' attitudes toward PND. The physician's age, gender, number of children, practice profile, and patients' socioeconomic level would all tend to colour a physician's experience, perceptions, and behaviour. Chapter 7 is devoted to the multivariate analysis of the impact of these factors.

Mother tongue, the indicator of where a person fits in the Canadian social structure, is another possible factor. In the Quebec/France study preceding this one, we were struck by the marked differences between Quebec's Anglophone and Francophone physicians (Renaud et al. 1991, 1992). Anglophones (about 20% of the Quebec medical profession) were more open to expanding the eligibility criteria for PND, less directive in their relationship with women and couples, and much more liberal on the topic of abortion. Since the mother tongue factor can be examined only through multivariate analysis in conjunction with province of practice (81% of Canada's Francophone physicians practise in Quebec), we will analyze its impact in Chapter 8. Contrary to what we thought before this study, Anglophones in Quebec are quite different from those elsewhere in Canada. In fact, they resemble Quebec Francophones more than their English-speaking colleagues in the rest of Canada.

In other studies, however, religious affiliation²⁴ and religious practice²⁵ are the most important determinants of the attitudes of both women and physicians toward PND (Sorenson 1973; Carlos and Cloutier 1976; Weitz 1979; Bernhardt and Bannerman 1982; Seals et al. 1985; Breslau 1987; Faden et al. 1987). Religion provides both precepts and a frame of reference. All religions have a certain view of death, abortion, and, at least indirectly, the means that could lead to these events. Obviously, the stronger is physicians' affiliation with their religious faith and the greater their activity within it, the higher is the probability they will adhere to its precepts and teachings. We will examine these factors in greater detail in

this chapter.

Attitude of Religions Toward PND

No religion formally disapproves of PND. Some avoid taking a position, and others are clearly in favour, but none advocates expanding the procedure. Churches mainly take positions on the ethical issues arising from developments in genetics, the chief one being selective abortion. In 14 of the 19 countries studied by Wertz and Fletcher (1989a), opposition by various religious groups was the main obstacle to the spread of PND. In order to prevent too large a number of selective abortions, the Norwegian government even set a specific quota on the number of amniocenteses that could be performed in a year (Royal Commission internal document).

Despite recent internal controversies, the Roman Catholic Church (and to a large extent the Orthodox Church) takes a very clear stand: PND must be condemned where it is done with the deliberate intention of terminating a pregnancy should the tests reveal an anomaly (Congregation for the Doctrine of the Faith 1987). For the Roman Catholic Church, a fetus is a human being from the moment of its conception, and the direct termination of fetal life, whatever the gestational age, is forbidden (Creighton 1986;

Fortin 1986). Furthermore, to the perplexing question of whether the mother or the child should be saved in the event of a difficult birth, the Catholic Church has long recommended saving the child.

At the opposite end of the spectrum is Judaism. With the exception of Orthodox Jews, who oppose abortion in any form and consider it murder, Jews have a liberal attitude toward abortion. In Judaism, the mother's life takes precedence over that of the fetus. The latter is even viewed as an "aggressor" if it threatens the life of the mother (Bleich 1981). Hence, the mother's anxiety or fears are sufficient grounds for requesting, and permitting, abortion. Therapeutic abortion is acceptable not because of the anomaly as such, but because of the physical and psychological burden that a disabled child would impose on the mother. It is seen as a private matter, to be considered in the light of Jewish law and the circumstances (Clévenot 1987; Longton 1987).

The various Protestant denominations hold intermediate positions. The Anglican Church, while never condemning abortion, asks the medical profession to show moral responsibility and discretion when dealing with a woman seeking an abortion. The United Church, while permitting abortion before the twentieth week of pregnancy, recommends that any such decision should take into account the fetus's right to life, the mother's right to a normal existence, and the family's right to a minimum standard of living. The positions of other Protestants (Presbyterians, Baptists, Lutherans, etc.) vary, falling between the positions of Catholics and Jews.

As for religions that we have termed "Oriental," Muslims reject abortion, considering it a "heinous crime," while Buddhists are more tolerant. However, when the life of the mother is in danger, both of these religions consider that she is the one who should decide.

We shall now examine the statistically significant differences that analysis has revealed between members of these various religions.

Use of Procedures

Ultrasound Scanning

Reasons

The use of ultrasound to screen for malformations was more acceptable to Catholic (69%) and Jewish (72%) physicians than to Protestant ones (47%) (A 6.1). This particular similarity between Catholics and Jews may no doubt be explained by the fact that 43% of Canadian physicians who are Catholic practise in Quebec, where a much more specialized medical profession has greater faith in ultrasound scanning as a prenatal diagnostic tool.

The use of ultrasound to screen for malformations was more acceptable to physicians who did not practise their religion (64%) than to those who did (55%). The latter were, at any rate, more reluctant to use PND (A 6.23).

Directiveness of Physicians

Faced with a woman who refuses ultrasound scanning, 74% of non-religious physicians — as opposed to their Jewish (58%), Oriental (58%), or Protestant (62%) counterparts — said they would accept the refusal as legitimate because the decision should be the woman's responsibility. On the other hand, physicians whose religious affiliation was Oriental (38%), Jewish (38%), or Protestant (32%) were the most likely to accept the refusal for the reason that the ultrasound scan is not important (A 6.2).

Amniocentesis and CVS

Appropriate Age of Access

Jewish physicians were most in favour of setting the eligibility threshold for amniocentesis at age 35 years (70%), and Protestants (other than members of the Anglican and United churches) the least in favour (57%) (A 6.3). A significantly larger number of the latter said they would raise eligibility to 40 years of age or reject the procedure. As regards CVS, Catholics, Protestants, and Jews were the most opposed to its use (20%, 18%, and 18% respectively) (A 6.4).

Physicians who practised their religion were generally more likely to want to raise the age of eligibility for amniocentesis beyond 35 (28%), and a greater number of them were also totally opposed to expanding the procedure (8%) when compared to their non-practising colleagues (2%) (A 6.24).

Eligibility Criteria

Jewish respondents differed from their colleagues in that a higher proportion of them said it was acceptable to allow a woman to have amniocentesis, even though she has no intention of aborting (47%, as opposed to 36% for Canada as a whole) (A 6.5).

Directiveness of Physicians

To a 36-year-old woman who had misgivings about amniocentesis because she feared spontaneous abortion, physicians who practised their religion were less likely (39%) to recommend going through with the procedure than those who practised their religion only occasionally (51%). Slightly more of the former would not recommend the procedure (19% versus 12% for those who practise occasionally and 13% for non-practising respondents) (A 6.25).

Jewish physicians were more ready to recommend amniocentesis to a 38-year-old primipara hesitant about the procedure (72% versus 63% for physicians as a whole).

Impact of Lawsuits on PND

Protestants were more sensitive than Catholics to the impact of lawsuits on PND (61% compared to 51%) (A 6.7).

Acceptability of Reproductive Methods for Circumventing Genetic Disorders

The use of artificial insemination or surrogate motherhood to prevent transmission of genetic disease was accepted more by non-religious physicians, 83% and 55% respectively, than by the Canadian medical profession as a whole (75% and 41%) (A 6.8, A 6.9). However, 85% of United Church physicians accepted artificial insemination, which is well above the overall score of 75%; with regard to surrogate motherhood, they were closer (43%) to the overall percentage.

Perception of Anomalies

The perception of the seriousness of certain conditions (paraplegia, sterility, behavioural problems, etc.) did not vary with religious practice. It did vary slightly, however, with religious affiliation (see Figure 6.1), Jewish physicians being more likely to regard as serious hypogonadism (35% compared to the Canadian mean of 22%), intellectual impairment (75% versus the Canadian mean of 61%), sterility (24% as opposed to 14% overall), cleft lip and palate (69% as against a mean of 49%), and lobster claw deformity (56% compared to 41%) (A 6.10).

Jewish respondents also said they would have difficulty accepting the idea of having a child with trisomy 21 (58% compared to 40% overall and 31% for Protestants) (A 6.11). The idea of having such a child was more accepted by physicians who practised their religion (51%) than by those who did not (28%) (A 6.27).

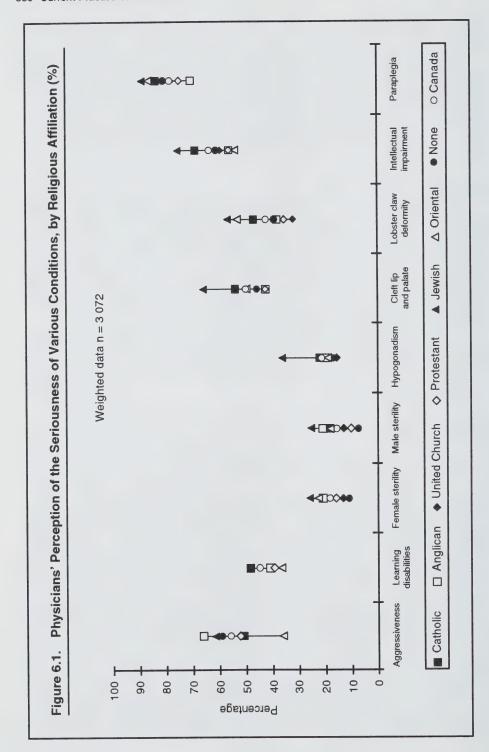
Social Choices

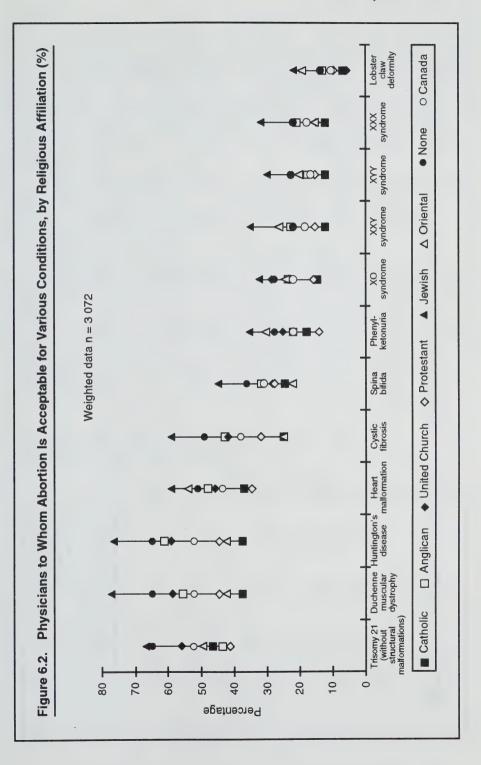
Attitudes Toward Abortion for Various Conditions

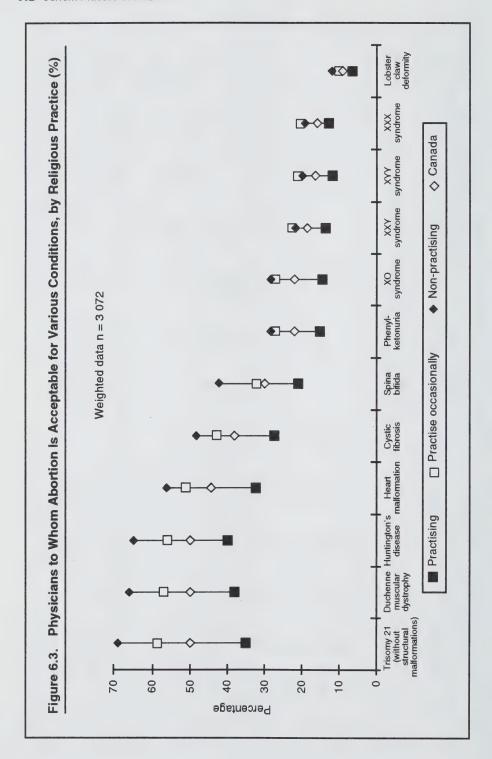
The most pronounced and consistent difference (see Figure 6.2) was between Catholic and Jewish physicians regarding the acceptability of abortion following PND. Jews were the most accepting of abortion, followed by non-religious respondents. Catholic physicians were the most strongly opposed, slightly more so than their Protestant colleagues. For example, aborting a fetus with Duchenne muscular dystrophy would be acceptable to 77% of Jewish physicians, but only to 38% of Catholic ones, while the idea of aborting a fetus with trisomy 21 (without structural malformations) was acceptable to 67% of Jews and 45% of Catholics (A 6.12).

Relatively fewer Jewish (40%) than Catholic (61%) respondents considered elective abortion less acceptable than selective abortion (A 6.13), and more of them (87%) rejected the idea that PND should be condemned when carried out with the deliberate intention of terminating a pregnancy (Catholics 63%, Protestants 64%) (A 6.14).

The acceptance of abortion among physicians who did not practise their religion was almost twice that of those who did (Figure 6.3). Of the conditions studied, the one that most justified abortion, according to







practising respondents, was Huntington's disease (39% in favour), followed by Duchenne muscular dystrophy (38%) and trisomy 21 (without malformations) (35%) (A 6.28). There was a consensus among physicians who did not practise their religion in favour of abortion if these conditions are detected (more than 60%). Preventing the birth of a potential schizophrenic, assuming that PND were possible, would only be acceptable to a minority: 11% of practising respondents and 20% of their non-practising colleagues (A 6.29). The latter were more in favour of discussing possible abortion with an alcoholic woman (40%) than were the former (24%) (A 6.31).

Slightly more than one-quarter of physicians practising their religion (27%) said that PND must be condemned if it is performed with the deliberate intention of terminating a pregnancy should the tests reveal an anomaly; one in 10 non-practising respondents concurred (10%) (A 6.32).

Directiveness of Physicians Regarding Abortion

As regards directiveness on the decision whether to abort (Tables 6.1 and 6.2), 71% of Protestant physicians and 77% of those practising Oriental religions said they believed that "a physician must be able to resist certain abortion demands when he or she considers an anomaly to be minor"; this was higher than Jewish physicians (48%) and Anglican and non-religious physicians (57%) (A 6.15). Protestant (39%) and Catholic and Oriental physicians (40%) were least in favour of parents' complete freedom of choice regarding abortion, while Jewish respondents were the most favourable (73%) (A 6.16).

Table 6.1. Physicians Agreeing with Various Statements on Directiveness, by Religious Affiliation (%)

	Cath.	Angl.	United Church	Prot.	Jewish	Oriental	None	CAN
A physician must be able to resist some abortion requests when he or she considers an anomaly to be minor (Q15 #2)	66	57	62	71	48	77	57	63
With respect to abortion, freedom of choice is an absolute	40	5 4						
(Q15 #4)	40	51	56	39	73	40	60	50

Table 6.2. Physicians Agreeing with Various Statements on Directiveness, by Religious Practice (%)

	Practising	Occasional	Non- practising	CAN
A physician must be able to resist some abortion requests when he or she considers an anomaly to be minor (Q15 #2)	70	60	53	63
With respect to abortion, freedom of choice is an absolute right (Q15 #4)	40	54	60	49

Clearly, physicians who practise their religion consider that part of their role is to resist certain abortion requests when they consider the anomaly to be minor (70% in favour), while their non-practising counterparts would be less directive (53%). The former were less inclined than the latter to say that parents' freedom of choice regarding abortion is an absolute right (40% compared to 60%) (A 6.33, A 6.34).

Disclosure of Information

Jewish physicians were more in favour (59%) than their Protestant colleagues (40%) or those belonging to an Oriental religion (31%) of disclosing information on the sex of the fetus in the case of early PND (A 6.17).

Perception of the Counterproductive Effects of PND

Jewish and non-religious physicians were less concerned about the potentially counterproductive effects of PND than were Catholics and Protestants. Only 34% of Jewish respondents were worried about "pathologizing" various conditions to a greater degree, as opposed to 56% for the other two groups (A 6.18, A 6.19). Regarding fears of increased intolerance, the percentage for Jewish practitioners remained at 34%, while those for Protestants and Catholics were 61% and 53% respectively.

Physicians practising their religion (59%) were much more anxious than their non-practising colleagues (40%) about effects of PND such as increased intolerance of the slightest fetal anomaly (59% versus 42%) or the "pathologizing" of previously normal conditions (58% versus 39%) (A 6.35, A 6.36).

Funding Priorities

Compared to Catholics (52%), physicians with an Oriental (75%) or Jewish (68%) religious affiliation attached great importance to evaluating the risk of exposure to mutagens and teratogens (A 6.20). The positions of

Jewish and Protestant respondents differed markedly on the acceptability of mass screening for cystic fibrosis (64% compared to 45%), as did their responses to the statement that PND cannot be considered a priority if one compares the percentage of birth defects to that of handicaps caused by social conditions: 31% of Jews agreed with the statement, compared to 52% of Protestants and an overall figure for Canadian physicians of 46% (A 6.21, A 6.22). Fifty-three percent of practising respondents agreed with the statement, as did 37% of non-practising respondents (A 6.38).

Summary

Generally speaking, non-religious and Jewish physicians were the most sympathetic to PND procedures, the most liberal about selective abortion, and the least directive in their relationship with couples if an anomaly was diagnosed. More of them tended to believe that amniocentesis should be available despite a woman's intention not to abort. They were less concerned about the potential counterproductive effects of PND. The position of Catholics was diametrically opposed, and very close to that of the denominations we have collectively termed "Protestant" (other than Anglican or United Church).

Regardless of their religious affiliation, those who did not practise their religion were always the most liberal, followed by those who practised occasionally and then by those who did so regularly. Tables 6.3 and 6.4 provide an overview of these sociocultural differences.

In short, as seen in the previous chapters, bivariate analysis tends to confirm the hypothesis that certain sociocultural characteristics determine physicians' attitudes toward PND. These characteristics include, in particular, "province of practice," "specialty," "religion," and "degree of religious practice." The second part of this study, which is devoted to multivariate analysis, describes the influence of these variables in greater detail.

Table 6.3. Significant Differences, by Religious Affiliation

	Catholic	Anglican	United Church	United Catholic Anglican Church Protestant Jewish Oriental None	Jewish	Oriental	None	Tot. pop. %
Ultrasound scanning No ultrasound should be done	1		+	+				50
Two ultrasounds should be done	+		+					16
Use ultrasound to screen for malformations	+			1	+			61
Use ultrasound to see fetus	+			ı				9
Accept refusal of ultrasound because decision is							-	09
Accept refusal of ultrasound because procedure				I			-	8
not important				+	+	+	1	25
Do not accept refusal and suggest seeing another								
	+		1					က
Ultrasound should not be subject to women's								
written consent	+	ı		i				29
Reliability of ultrasound scanning								
Confidence in ultrasound to detect:								
- limb malformation							ı	42
- hydrocephaly					+			45
- trisomy 21				+	+		1	က
Eligibility criteria for amniocentesis and CVS								
Accept age 35 as eligibility threshold			+	ı				62
Amniocentesis should not be available	+			+				52
Set eligibility threshold for CVS at age 35	ı	+	+		+			52
CVS should not be available	+		ļ	+	+			<u>0</u>
Make amniocentesis available without criteria if			-					20
patient pays	ı		+					5

Cat	tholic /	Anglican	United	United Catholic Anglican Church Protestant Jewish Oriental None	Jewish	Oriental	None	_
Funding priorities								
PND is not a priority if one compares the								
percentage of birth defects to that of handicaps of								
social origin				+	ı		ı	
Attach great importance to evaluating risk of								
exposure to mutagens or teratogens during								
pregnancy –			+					
Favourable to mass screening for cystic fibrosis if								
all carriers could be identified			+	1	+			
Only statistically significant differences are indicated.								
Tot. pop. = percentage of physicians in favour for Canada as a whole.	ınada as	a whole.						
+ = above Canadian percentage.								
 = below Canadian percentage. 								

46 60 52

Table 6.4. Significant Differences, by Religious Practice

	Practising	Practise occasionally	Non- practising	Tot. pop
Ultrasound scanning Use ultrasound to screen for malformations	+		-	61
Eligibility criteria for amniocentesis				
Never perform amniocentesis Recommend procedure to a	+		-	5
reluctant 36-year-old woman Recommend procedure to a	-	+		44
38-year-old woman	-	+		62
Impact of lawsuits There is a danger PND will be used more often than medically required because				
of threat of lawsuits	+			56
Acceptability of abortion Abortion is acceptable for: - trisomy 21 (without				
structural malformations) - Duchenne muscular	-		+	51
dystrophy	-		+	51
- Huntington's disease			+	51
severe heart malformations	-		+	43
- cystic fibrosis	-		+	37
- spina bifida	-	+	+	30
- phenylketonuria	-	+	+	21
- Turner's syndrome	-	+	+	21
- Klinefelter's syndrome	-	+		17 16
- XYY syndrome		+		16
- XXX syndrome	-	+		16
- schizophrenia	-			16
Would agree to have a child				
with trisomy 21	+		-	71
Discuss abortion with				
alcoholic women	-		+	31
Condemn PND if done with intent to abort	+		-	19

conditions previously considered normal

Funding priorities
PND is not a priority if one
compares the percentage of
birth defects to that of
handicaps of social origin

abnormality

PND increases intolerance of

Table 6.4. (cont'd)				
	Practising	Practise occasionally	Non- practising	Tot. pop
Directiveness with regard to abortion Parents have an absolute right to freedom of choice concerning abortion Physician must resist certain abortion requests	-+		+ -	49 63
Perception of the counterproductive effects of PND PND makes disorders out of				

51

50

47

Only statistically significant differences are indicated.

Tot. pop. = percentage of physicians in favour for Canada as a whole.

+ = above Canadian percentage.

= below Canadian percentage.

Part 2. Multivariate Analysis

Chapter 7. Influence of Sociocultural and Professional Characteristics

In this chapter, we will examine the influence of various sociocultural and professional factors on physicians' attitudes toward PND technology and selective abortion. Unlike the bivariate analyses in the previous chapters, the multivariate analyses will make it possible to examine several predictive variables simultaneously. The combined effect of several factors

can thus be analyzed, as well as the net influence of each when the others are held constant. These analyses will help integrate the findings on physicians' attitudes toward PND techniques and the ethical problems they raise, and thus facilitate the development of a model of the sociocultural determinants of attitudes toward the various aspects of PND.

The three main lines of inquiry are, again, attitudes toward PND procedures, perception of anomalies, and questions pertaining to social choices. However, given the complexity of the questions raised, the analysis focusses on a few specific issues within each area. For procedures, the focus is on the eligibility criteria for amniocentesis and CVS (the scale measuring the willingness to expand access to amniocentesis and CVS), the tendency to favour testing for predisposition, and the issue of funding priorities. For anomalies, physicians' attitudes toward the seriousness of the anomalies ("perception of seriousness of anomalies" scale) was examined. For social choices, attitudes toward the acceptability of selective abortion ("acceptability of abortion" scale) were studied, as well as physicians' directiveness with regard to the abortion decision ("directiveness" scale).

Since these themes are in fact different aspects of the overall issue, the analyses take these inter-relations into account, assuming a cause and effect relationship between the various scales.26 We assumed that physicians' attitudes toward the use and development of PND procedures can be influenced by their attitudes toward certain ethical dilemmas, in particular that of selective abortion. Attitudes toward selective abortion can also be determined by perceptions of the gravity of various types of impairment or anomaly. The acceptability of abortion can influence a physician's directiveness with regard to the decision whether to abort. We have assumed that the various attitudes toward PND are related as follows: perception of seriousness \rightarrow acceptability of abortion \rightarrow directiveness with respect to the abortion decision \rightarrow attitudes toward the use and development of PND procedures. The analyses will be presented in this order. Before reporting our findings, however, we will describe the manner in which the various scales have been established and validated, and the methods of multivariate analysis used.

Establishing and Validating the Scales

The principle involved in establishing a scale is to combine answers to various questions that measure a common concept. For a scale to be statistically meaningful and useful, its various elements must be sufficiently correlated and shown to measure the same thing. Cronbach's alpha (Cronbach 1951) assured us that our scales were internally consistent. Cronbach's alpha is a function of the number of items and the mean inter-item correlation. It varies between 0 and 1; the higher it is, the greater the internal consistency of the scale.

"Perception of Seriousness of Anomalies" Scale

Questions 9 and 10 were used to establish this scale. In question 9, respondents were asked to evaluate the seriousness of three sex chromosomal anomalies (XYY syndrome, Klinefelter's syndrome, and Turner's syndrome). A brief description of the three anomalies followed the questions. The answers were given on a five-point Likert scale ranging from "not at all severe" to "extremely severe." In question 10, respondents were asked how difficult they would find it to be the parents of a child with certain problems. The answers were also given on a five-point Likert scale ranging from "not at all difficult" to "very difficult."

Scores were based on the mean value of each individual's answers to the 13 items of the two questions. Each mean value was then multiplied by 13. This prevented respondents who failed to answer some questions from obtaining a lower score. Scores ranged from 13 to 65 (alpha = 0.84) in order of increasing perceived seriousness. This scale measures physicians' general attitude on the gravity of anomalies. The higher the score, the greater the perceived gravity.

"Acceptability of Abortion" Scale

The "Acceptability of Abortion" scale was established on the basis of answers to the items in question 11 on the acceptability of abortion for various anomalies. The question was worded as follows: "For each of the following conditions diagnosed in a fetus, please indicate the extent to which you believe pregnancy termination is acceptable." All the items except the one on aborting a fetus of the undesired sex were included in the scale. The answers were entered on a five-point Likert scale ranging from "not at all acceptable" to "totally acceptable."

Like the previous one, this scale was computed by taking the mean value of the 12 items for each individual and multiplying it by 12. The scale ranged from 12 to 60 (alpha = 0.95), 12 referring to individuals categorically opposed to abortion, and 60 to those who are very favourable to it, for each of the 12 conditions given.

"Directiveness on Abortion" Scale

This scale was prepared by adding the answers to three statements:

Q15 **Statement 2**: A physician must be able to resist some abortion requests when he or she considers an anomaly to be minor.

Statement 4: With respect to abortion, parents have an absolute right to freedom of choice.

Statement 6: Physicians, not parents, should decide which fetal anomalies warrant pregnancy termination.

Answers ranged from "totally disagree" to "totally agree" on a five-point Likert scale. Coding for statement 4 was reversed (1 = 5, 5 = 1, etc.) to remain consistent with the other two statements. As with the other two scales, the mean value of each individual's answers to the three statements was computed and multiplied by three. This procedure was used for

respondents who had answered at least two of the three statements. Cronbach's alpha was 0.54. The scale ranged from 3 to 15, from least directive to most directive. "Directiveness" on this scale refers to the notion of restricting access to abortion. Thus, a physician who scored high on this scale was one who tended to restrict access to abortion.

"Expanded Access to Amniocentesis and CVS" Scale

In order to measure physicians' willingness to expand access to PND, we set up an indicator based on the various questionnaire items where respondents had to indicate whether or not they would order the test and, if so, at what age. Although necessarily somewhat arbitrary, this indicator gives a general idea of a physician's willingness to provide broader access to the more invasive PND techniques.

The following six questions were used:

- Q5: "Irrespective of present policies, at what age do you think antenatal screening for fetal chromosome anomalies should be offered to women who have no family history of problems?"

 (A) Amniocentesis (at all ages, <30, 30, 31, 32, 33, 34, 35, 36, 37, 38, 39, ≥40, never)
 - (B) CVS (at all ages, <30, 30, 31, 32, 33, 34, 35, 36, 37, 38, 39, ≥40, never)
- Q8.1: "A couple is very worried about giving birth to an abnormal child. The woman says she has insomnia and frequent nausea. She is 33 years old and has no history of genetic disorders, but feels very anxious because a 30-year-old friend recently gave birth to a child with trisomy 21. They request amniocentesis." (answers on a scale from 1 to 5, "Not at all acceptable" to "Totally acceptable")
- Q8.3: "A 33-year-old woman requests amniocentesis. She is knowledgeable about the risks of the test, and agrees that at her age the risks of having a child with trisomy 21 are rather small. However, since prenatal tests exist, she believes it is for her to decide whether to have the test, and to choose the risks she is willing to take." (answers on a scale from 1 to 5, "Not at all acceptable" to "Totally acceptable")
- Q15, dilemma 5: "A woman who does not meet general policy guidelines for an amniocentesis should have access to the test if she is willing to pay the costs of this service herself." (answers on a scale from 1 to 5, "Not at all acceptable" to "Totally acceptable")
- Q15, dilemma 27: "Amniocentesis should be made available to all women, no matter their age, or marital or socioeconomic status." (answers on a scale from 1 to 5, "Not at all acceptable" to "Totally acceptable")

As for previous scales, a mean value was computed for the six variables and was multiplied by the number of variables making up the

score (alpha = 0.68). A score was computed for all individuals who answered at least four of the six questions. As all answers did not have the same value range (two ranged over 13 points, the other four over 5 points), each answer was weighted so that it would have equal weight in computing the score. The higher a physician's score on the scale, the more willing he or she was to expand access to PND procedures by altering the age threshold or criteria.

Method of Analysis

The influence of sociocultural and professional factors on the various scales was analyzed through variance analysis and multiple classification analysis (MCA). These methods allowed the simultaneous impact of various predictive variables on each of the scales to be evaluated. Variance analysis identifies the variables that have a significant impact on a scale, and MCA is used to evaluate the direction and intensity of that impact. MCA is based on the additive model assumption that the position of each individual on a given scale is the result of the addition of the effects associated with the individual characteristics measured. MCA and the variance analysis that accompanies it²⁹ have the advantage of not requiring that predictors be independent. This is obviously of prime importance since our predictors, being the result of a survey, cannot be independent, unlike the experimental model where the subcategories of all variables have an equivalent number of subjects.

MCA is very similar to multiple regression analysis. It provides, as does regression, a multiple correlation coefficient (R) and a determination coefficient (R²), which make it possible to evaluate the combined influence of multiple predictors on each scale. Unlike regression, however, MCA makes it possible to clearly express the direction and intensity of the effects of nominal predictors as adjusted deviations. It measures the effect of a subcategory of a predictor expressed as a deviation from the overall mean where other predictors are held constant. In addition, for each predictor, MCA provides a beta coefficient that is similar to the "b" of regression, but applies to nominal predictors. This beta is a measurement of the net influence of each predictor (combining all subcategories) on the dependent variable

The effects of the interaction between two or more predictors on the dependent variable cannot be evaluated with MCA, however. It does not show whether the influence of the medical specialty or ethnic origin on abortion acceptance is the same between provinces or for men and women. To offset this limitation, variance analyses were done on each pair of predictors and for every scale to identify significant interactions. We had to limit the interaction analyses to predictors taken two at a time, primarily because of the small number of respondents in some categories, but also because of the large number of categories for some variables. We were thus able to identify "ecological" variables, that is, variables that interact with

several others. Such variables seem to indicate that the main effects uncovered by MCA and overall variance analysis are not the same for each level of those predictors. In fact, the "province" variable is the only one that exhibited interaction with several variables, and this was true for every scale. For example, with regard to the perception of the seriousness of various anomalies, interactions have shown that "distance from a genetics centre," "religion," "socioeconomic background of a physician's clientele," and "ethnic origin" do not have the same influence from one province to the next. These interactions led us to conduct variance analyses and MCAs for each province and to take a close look at how the influence of sociocultural factors differs from one province to the next.

Analysis of "Perception of Seriousness of Anomalies"

Figure 7.1 shows the score distribution on the "perception of seriousness of anomalies" scale. Scores ranged from 13 to 65 with a mean value of 41.1. If scores were reset on a 1 to 5 scale, the mean value would be 3.2, which means that, as a group, Canadian physicians considered the anomalies as a whole moderately serious. However, as can be seen from the spread, this perception varies greatly from one physician to the next.

Table 7.1 presents the results of variance analysis for our Canadian physicians as a group. The seven variables presented are the only ones that proved to be significantly linked to the "perception of seriousness" scale. They were province, ethnic origin, religion, medical specialty, number of children, medical school attended, and distance from a genetics centre. The relationships between these variables and the "perception of seriousness" measurements were significant with a minimal probability of Type I³⁰ error. However, judging by the percentage of the variance that they account for, these relationships are not very strong. In fact, the variables account for only 10% of the variance in the "perception of seriousness" scale. Province alone accounted for about 1.3% of the variance; at thic origin 1.6%; religion 1.4%; medical specialty, number of children, medical school attended, and distance from a genetics centre, 1% each. These combined variables accounted for another 2% of the variance.

Table 7.2 presents, for Canada as a whole and for each of the provinces, the betas and adjusted deviations from the mean (d') for each of the predictors having a significant bearing (p < 0.01) on the perception of seriousness. For example, Ontario's Catholics are 1.4 points from the mean (40.3) on a scale ranging from 13 to 65, controlling for other factors. The beta for religion in Ontario is 0.18, which indicates religion has less influence in Ontario than in British Columbia (beta = 0.22).

The effects identified in the overall Canadian sample did not apply provincially, except for Ontario, where the significant effects identified, and the magnitude of the adjusted deviations, very closely matched the Canadian sample. Analysis by province is all the more revealing because it identifies the trends that can be observed in several provinces.

Figure 7.1. Score Distribution on "Perception of Seriousness" Scale Mid-point Frequencies 14.0 | 16.5 l 4 19.0 I* 8 21.5 I 14 24.0 | **□ 30 26.5 | **** 56 29.0 | ******* 103 31.5 | ********* 139 34.0 | ***************** 308 36.5 | ******************* 253 39.0 | ******* 507 41.5 | ******************* 310 44.0 | ****** 471 46.5 | ************** 237 49.0 | ************* 271 51.5 | ******* 103 54.0 | ****** 107 56.5 | ***□ 38 59.0 | * 25 7 61.5 l 64.0 | * 12 +----+---+---+---+---+----+----0 120 240 360 480 600 **Descriptive statistics** 7.470 Maximum 65 3 006 Standard deviation Mean 41.070 13 Missing values 66 Minimum

Source of variance	Sum of squares	DF	Mean squares	F	Sig. of F
Main effects	14 884.263	26	572.472	11.539	0.000
Ethnic origin	2 402.123	5	480.425	9.683	0.000
Province	2 036.627	6	339.438	6.842	0.000
Religion	2 075.586	7	296.512	5.977	0.000
Number of children	1 223.730	3	407.910	8.222	0.000
Medical specialty	1 414.090	3	471.363	9.501	0.000
Distance from a genetics					
centre	1 158.970	1	1 158.970	23.360	0.000
Where attended medical					
school	1 321.204	1	1 321.204	26.630	0.000
Accounted for	14 884.263	26	572.472	11.539	0.000
Residual	131 557.787	2 652	49.613		
Total	146 442.050	2 678	54.683		

n = 3072.

Number of missing values: 393 cases (13%).

Although the variance in the perceived seriousness scores was comparable from province to province, there were sizable differences between provinces in the percentage of the variance accounted for by sociocultural and professional factors. While the percentage of variance accounted for by sociocultural and professional factors was between 10% and 15% for Canada as a whole and most provinces, it reached 21% in Saskatchewan and was only 2% for Quebec. This is an important finding. since these percentages of accounted-for variance indicate the magnitude of the differences within each province or, conversely, the tendency of physicians to conform to the group to which they belong (religious, ethnic, The higher percentage of accounted-for variance in Saskatchewan may be explained in part by the smaller size of the sample, but a similar explanation does not hold for Quebec. A higher proportion of variance was accounted for in provinces of similarly large size, such as Ontario. Sociocultural factors, therefore, had very little bearing on the Canadian physicians' perceptions of the seriousness of anomalies. They had practically no influence on Quebec physicians.

This was all the more surprising since Quebec physicians clearly differ from other Canadian physicians in that they perceive anomalies overall as much more serious. The perceptions of other Canadian physicians are very consistent in that regard. As noted in Chapter 4, Quebec physicians, especially those of French origin, consider most individual anomalies as being more serious than do their counterparts in other provinces. Quebec's

Canada Atlantic Qu	Canada	Atlantic	Quebec	Ontario	Manitoba	Sask.	Alberta	BC/NWT
n¹ Means Standard deviations R²	3 072 41.1 7.5 0.10	240 39.9 7.3 0.11	738 44.7 7.1 0.02	1 249 40.3 7.7 0.10	152 40.9 6.8 0.11	88 41.2 7.1 0.21	253 40.0 7.6 0.08	353 40.9 6.7 0.13
Provinces Ethnic origin British French Asian Jewish European	0.18 0.17 0.17 1.1- 1.4 1.7*	-0.5	0.14 0.3 0.3 0.3 1.1	0.8 0.13 0.7 0.7 0.7 0.5 0.5	-0.8	0.1	0.0 0.20 0.09 0.07 0.07 0.07 0.03	0.1
Religion Catholic Anglican United Church Protestant Jewish Oriental	0.15 0.07 0.7 0.7 0.7 1.1.1 *1.1.0	0.20 -0.6 -1.7 -1.5		0.18 0.14 0.04 0.03 0.03 0.03 0.04 0.04 0.04 0.0				2.1.2 -2.1.3 -1.6 -1.3 -0.2
Specialties GP Obstetrician Paediatrician Radiologist	0.10 0.3 0.3 -0.8 1.4 1.4	8. 4. F.	0.8 4.1.1 4.1.4	0.15 0.3 -0.8 -1.7 2.9	0.30 1.30 1.30 1.30 1.30	0.5.1 0.5.0 0.0.0	6. 4. 6. 6. 6. 6. 6. 6. 6. 6. 6. 6. 6. 6. 6.	6.5 9.6 9.6 9.6
Number of children None 1 child 2 children	0.09	1.0 1.1 0.3	1.0	0.10 0.3 1.6 0.6	0.3 1.6 0.6	0.35 4.6 4.6	4.6 4.6 1.5	4.6 4.6 1.5

		0.7 0.7 -1.8	0.13 -0.5 1.6			0.17 3.1 0.3 -0.5 -0.9 -1.0	
			0.13 -0.6 1.6		0.18 -0.6 3.3		
_				0.35 -3.5 2.2 1.4			
-		0.29 1.5 -2.8					
0.12 -0.5 1.9		0.09 0.4 -1.2			•		justed deviations
			0.07 -0.2 0.9				nted. ted and non-ad
	0.25 1.9 -1.9						e is not weig etween adjus
0.11 -0.4 1.7	0.10 0.5 -1.0						ians per provinc e discrepancy b
Med. school attended Canada Elsewhere	Distance from a centre < 100 km > 100 km	Area Urban Rural	Gender Male Female	Early convert (to technology) No Average Yes	Clientele Middle-class Underprivileged	Age Under 35 35 to 39 years 40 to 49 years 50 to 59 years 60 years or +	 Number of physicians per province is not weighted. Indicates a sizable discrepancy between adjusted and non-adjusted deviations. Betas are in bold.

English physicians, with an adjusted deviation from the mean of -2.8, were closer to the Canadian mean. It was also surprising to see that wherever ethnic origin had a bearing on perceived seriousness, *physicians of French origin considered anomalies as most serious*. Apart from Quebec and Canada as a whole, this applied notably to Ontario physicians and even more so to those from Alberta. At the opposite end of the scale, physicians of British origin (including "English Canadians") were those who considered anomalies least serious.

The influence of religion on the perception of anomalies may largely be attributed to the different views held by Jewish physicians and their Catholic and Protestant counterparts. Jewish respondents considered the various anomalous conditions as much more serious than did physicians of the other religions, more particularly Catholics and Protestants, who were at the opposite end of the spectrum.

Medical specialty did not seem to have any great bearing on the manner in which physicians perceived anomalies. This factor had a significant influence only in Ontario and Manitoba, and the observed differences showed no systematic trend.

However, differences linked to the number of respondents' children did suggest a trend. Looking at the sample as a whole, and Ontario and Saskatchewan in particular, we observed that physicians with three or more children considered anomalies overall as less serious. In Saskatchewan, this factor was particularly marked and led to wider discrepancies.

Although the perception of seriousness was significantly linked in the Canadian sample to the place where a respondent had studied medicine and to the distance from a genetics centre, each of these factors had significant influence in only one region: Ontario, for the place of study, and the Atlantic provinces, for the distance from a genetics centre. It is therefore difficult to determine the influence of these factors in anything other than a provincial context. The same was true for a physician's age and the socioeconomic background of the clientele, which each played a role in only one province (British Columbia in the first case, and Alberta in the second).

Lastly, although they were not significant for the Canadian sample, two factors seemed to have a marked influence on a number of physicians: practice area and gender. In Ontario, Manitoba, and British Columbia, physicians in urban areas perceived anomalies as more serious than their colleagues in rural practice. Female respondents perceived anomalies as being more serious than did male respondents.

In summary, although physicians agreed on the relative seriousness of various fetal anomalies, there was considerable variation in the way they perceived the seriousness of anomalies as a whole. Such sociocultural factors as province or region of practice, religion and ethnic origin, number of children, practice area, and gender nonetheless had an influence on their perceptions. We noted that Quebec physicians, especially Francophones,

considered anomalies overall as more serious than their Canadian colleagues. The same was true of physicians of French or Jewish origin in several Canadian provinces. Conversely, male physicians, physicians with several children, and those with a rural practice tended to see anomalies overall as less serious. Yet, while these sociocultural factors may account for a small part of the variance, the major portion remains unaccounted for, and is seemingly attributable to individual differences alone.

Analysis of "Acceptability of Abortion"

Figure 7.2 shows the distribution of scores on the "acceptability of abortion" scale. A very large number of individuals scored at the bottom of this scale. These respondents, who made up 14% of the sample (n = 428). answered that abortion was unacceptable for any anomalous condition. The proportion of such respondents among GPs (17%) was almost double that observed among other physicians (9%). These respondents were found in larger numbers among Catholics (23%) and Protestants (26%), and they were more likely to practise their religion than were other physicians. They were largely of British and European origin and were more likely to describe themselves as conservative than were other respondents. There were far fewer respondents of this type in Quebec (5%) than in other provinces, and far more in Saskatchewan (32%). Despite these differences, we thought it was important to include these physicians in the multivariate analyses. The F test on which variance analysis and MCA are based is very effective in dealing with this type of deviation from normal score distribution. It is therefore preferable to include all individuals in the group for the purpose of analysis. For verification purposes, the results obtained by excluding these individuals from the analyses were, in fact, very similar to those reported here, which were obtained by including them.

Scores on the "acceptability of abortion" scale ranged from 12 to 60 with a mean value of 31.4. If this mean value is divided by the number of items making up the scale, a mean value of 2.6 on a five-point scale is obtained, which means that physicians were, on the whole, slightly opposed to abortion for the conditions listed overall. The mean shifted very little (2.95 out of 5) if respondents fundamentally opposed to abortion were excluded. However, as the score distribution indicates, there was also a substantial divergence in the acceptability of abortion for these conditions, since clinical seriousness varied greatly.

Variance analysis identified eight factors significantly associated with the "acceptability of abortion" scale among Canadian physicians as a group. These factors accounted for 25% of the variance in the "acceptability of abortion" scale. If perceived seriousness is added as a co-factor to these variables, 33% of the variance is accounted for. We believed it was important to include this variable as a co-factor in the analysis because of its strong association with the acceptability scale (r = 0.36), and in order to obtain the direct effects of the predictors on the acceptability scale — i.e., the effect of the predictors on the "acceptability of abortion" scale when the

Figure 7.2. Score Distribution on "Acceptability of Abortion" Scale

Frequencies	Mid-point
428	11.0 ***********************************
66	13.5 ******* □
122	16.0 *********
82	18.5 I *******
154	21.0 ************
110	23.5 *********
181	26.0 **************
150	28.5 *************
245	31.0 ***********************************
183	33.5 **************
314	36.0 ***********************************
157	38.5 *************
231	41.0 ***********************************
118	43.5 ********** □
172	46.0 *************
84	48.5 ******* ⁻
84	51.0 *******
43	53.5 **** 🗆
58	56.0 *****
27	58.5 **
41	61.0 ***
	0 100 200 300 400 500
Descriptive s	statistics 3 049 Standard deviation 12.983 Maximum 60
	1.429 Minimum 12 Missing values 22

"perception of seriousness" is constant. The effects of the variables on the acceptability scale remained essentially the same with the addition of this co-factor, which indicates that the perception of seriousness has an independent influence on abortion acceptability. The results of variance analysis on the Canadian sample are presented in Table 7.3. All effects were significant, with a very low probability of Type I error. The F of the cofactor is particularly large, which confirms the strong correlation between the "acceptability of abortion" scale and the "perception of seriousness" The two main factors were religious commitment (practising, occasional, non-practising: 4.2% of the variance) and religion (4.9%), followed by medical specialty (2.1%), province (1.1%), ethnic origin, number of children, and age group, each of which accounted for 1% of the variance. The results of MCA on the Canadian sample and on each of the provinces are presented in Table 7.4. Only the betas and adjusted deviations of predictors with a significant influence on abortion acceptability are reported.

What is striking in this table is the consistency of the results. Predictors with a substantial influence for Canadian physicians as a group had a comparable influence in several provinces. This was particularly true for religious practice, religion, medical specialty, and ethnic origin. All these factors show a consistent influence on physicians' attitudes toward selective abortion. These factors, except for religious practice, also had an influence on physicians' perceptions of the seriousness of anomalies. Here, however, the influence of these factors appeared to be felt more strongly and by a larger number of respondents. This was apparent from the number of provinces where these factors played a significant role, and particularly from the magnitude and consistency of the observed discrepancies.

The percentages of variance accounted for by the R² factors at the top of the table indicate that sociocultural and professional factors as a whole accounted for a larger part of the variance in the "acceptability of abortion" scale than in the "perception of seriousness" scale. While percentages of variance accounted for were in the 10% range in the latter case, for the abortion scale they are in the 25% range and even exceed 50% in the case of Alberta. Hence it appears that the acceptability of abortion is largely influenced by sociocultural factors. If one adds "perception of seriousness" to those factors, 30% of the variance in "acceptability of abortion" is accounted for — nearly 60% in Alberta. In fact, "perception of seriousness" accounted for 6%-7% of the variance in the "acceptability of abortion" scale, nearly 20% in the Maritimes — hence the very strong relationship between these two attitudes. Thus, the more physicians consider anomalies overall to be serious, the more they will be favourable to selective abortion. This coincides with what we observed in the bivariate analyses, where we saw that those anomalies that physicians considered serious were also the ones for which a larger number of physicians would find aborting a fetus to be justified.

Table 7.3. Variance Analysis of Acceptability of Abortion Scale

Source of variance	Sum of squares	DF	Mean squares	F	Sig. of F
Covariates Perception of	50 969.106	1	50 969.106	441.292	0.000
seriousness	50 969.106	1	50 969.106	441.292	0.000
Main effects	93 079.345	30	3 102.645	26.863	0.000
Religion	21 712.950	7	3 101.850	26.856	0.000
Religious practice	18 671.788	2	9 335.894	80.830	0.000
Specialty	9 075.862	3	3 025.287	26.193	0.000
Ethnic origin	3 612.692	5	722.538	6.256	0.000
Province	4 709.164	6	784.861	6.795	0.000
Age groups	3 368.963	4	842.241	7.292	0.000
Number of children	3 237.898	3	1 079.299	9.345	0.000
Accounted for	144 048.451	- 31	4 646.724	40.231	0.000
Residual	296 611.194	2 568	115.500		
Total	440 659.645	2 599	169.550		

n = 3072.

Number of missing values: 472 cases (15%).

The systematic nature of the influences observed from one province to the next leads to an interesting interpretation of the interaction between "province" and the other sociocultural factors with regard to abortion acceptability. It can be seen that a large part of these interactions are due to subgroups of physicians, which react very differently depending on their province. For example, the attitude of Protestants is known to differ markedly from one province to the next. In other words, there was an interaction between religion and province, although overall trends emerged for Catholics and Jews.

Apart from the fact that the predictors operated somewhat differently from one province to the next, "province" had a determining influence on abortion acceptability. This is apparent from the analysis of the Canadian sample: with all sociocultural factors controlled for, "province" accounted for a significant proportion of the variance in abortion acceptability. Three groups can be discerned from the adjusted deviations. All other things being equal, ³² Quebec and British Columbia (with deviations of 1.3 and 1.2 respectively) were the provinces most favourable to abortion. If all other predictors are not controlled for — that is, if physicians' attitudes are presented as they are in their respective provinces — then Quebec stands out even more, with a mean value of 35.4 versus 31.4 for Canada as a whole (32.4 for British Columbia). Saskatchewan (d' = -5.1) and Manitoba

ions for Each of the Predictors with a Significant Bearing on Acceptability of	la and for Each Province
Adjusted Deviations for Each	For All of Canada and for Eacl
Table 7.4. ₽	Abortion:

-0.8 1.3 -0.2 -2.0 -5.1 0.2 0.19 0.25 0.22 0.23 0.35 0.36 -1.8 -4.4 -2.8 -2.2 -2.6 -4.8 3.7 0.3 1.6 1.0 7.4 2.0 -1.2* 3.2 3.7 5.0 3.9 6.9 -1.2* 3.2 3.7 5.0 3.9 6.9 -0.26 0.20 0.29 0.47 0.21 -6.2 -1.1 -6.4 -4.8 -4.5 -4.5 -6.2 -1.1 -6.4 -4.8 -4.5 -0.9 2.3 - 1.1 5.5 -4.5 -0.9 2.3 - -1.1 -6.7 0.8* -7.5 - - - - - - - 3.2 4.8 2.5 4.8 - - - 3.2 4.8 2.5 4.8 - - - - - - - - -	n Mean Standard deviations R² (factors) R² (gravity)	Canada 3 072 31.4 13.0 0.33 0.25 0.08	Atlantic 240 29.7 12.8 0.31 0.12	Quebec 738 35.4 11.6 0.26 0.19	Ontario 1 249 31.3 12.7 0.30 0.23 0.07	Manitoba 152 28.7 12.3 0.39 0.34	Sask. 88 23.7 11.5 0.22 0.16	Alberta 253 29.7 13.6 0.57 0.06	353 32.9 13.4 0.42 0.36
0.26 0.29 0.47 0.21 -6.2 -1.1 -6.4 -4.8 -4.5 0.5* - 1.3 7.5 -0.9 2.3 - 1.1 5.5 4.7 0.1 4.7 1.7 -6.7 0.8* - - - - - 3.2 4.8 2.5 4.8 1.3* 0.17 0.20		0.11 0.23 -3.3 2.1 3.1	0.19 0.19 -1.8 3.7 -1.2*	1.3 0.25 -4.4 0.3 3.2	-0.2 0.22 -2.8 1.6 3.7	-2.0 0.23 -2.2 1.0 5.0	-5.1 0.35 -2.6 7.4 3.9	0.2 0.36 -4.8 2.0 6.9	0.18 0.18 3.4 0.2
		0.29 9.55 9.00 0.4* 0.6 2.6 0.6	0.26 -6.2 0.5* 0.1 3.2	0.20 -1.1 4.7 5.9 	0.29 -6.4 1.3 1.1 1.7 -0.6 2.5	6.7 7.5 7.5 6.7 6.0 6.0 7.0 6.0		0.21 -0.9 -0.9 4.7 0.8*	0.35 -9.4 -9.4 -0.8* -0.8* -0.8*

n Mean Standard deviations R² (factors) R² (gravity) Obstetrician Paediatrician Radiologist	Canada	200						
viations In an		Atlantic	Quebec	Ontario	Manitoba	Sask.	Alberta	BC/NWT
viations In an	3 072		738	1 249	152	88	253	353
viations tn an	31.4		35.4	31.3	28.7	23.7	29.7	32.9
t an	13.0		11.6	12.7	12.3	11.5	13.6	13.4
un an	0.33		0.26	0.30	0.39	0.22	0.57	0.42
ın an t	0.25		0.19	0.23	0.34	0.16	0.51	0.36
-	0.08	0.19	0.07	0.07	0.05	90.0	90.0	90.0
Paediatrician Radiologist	4.5		3.2	5.2				8.5
Radiologist	0.8		8.0-	0.7				4.4
	2.4		2.3	1.7				7.5
Ethnic origin	0.13		0.21	0.11			0.28	0.23
British	-0.3		4.9	0.1			-0.2	0.0
French	3.0		-1.1	3.7*			14.1	
Asian	0.1		-0.5	-0.8			2.9	2.3*
Jewish	-0.6*		6.2	2.9*			1	,
European	-2.9			-2.3			-5.4	-5.9
East European	0.4			0.2			6.0	5.8*
Number of children	0.10		0.14				0.17	
None	1.5		1.6				2.2	
1 child	2.8		3.9				6.8	
2 children	9.0		-0.1				0.2	
3 children or +	-1.2		-1.4				-2.0	
Age	0.10						0.24	
Under 35	-3.0						-7.4	
35 to 39 years	-0.4						0.4	
40 to 49 years	0.5						0.8	
50 to 59 years	1 .3						4.3	
60 years or +	1.4						4.3	1000

Well-to-do 3.3 -0.2 2.3 -0.7 Underprivileged -1.7 -4.4 5.1 Vixed - - - Alixed - - - stance from a stance from a entre 0.20 - c 100 km 2.1 - > 100 km -3.6	ss -0.2 2.3 2.3 eged -1.7 7.9 -4.4 7.9 c.20 2.1 a sizable discrepancy between the adjusted and non-adjusted deviations.	Clientele	0.14	0.28		0.14
e-class	e-class — 0.2 2.3 — 4.4 1.7 7.9 — 4.4 7.9 — 5.2 — 4.4 7.9 — 5.2 — 4.4 7.9 — 5.0 — 5.1 — 5.6 — 5.	Well-to-do	3.3			ı
rprivileged –1.7 –4.4 7.9 –0.20 ce from a	rprivileged — 1.7 T.9 ce from a Name of physicians per province is not weighted. cates a sizable discrepancy between the adjusted and non-adjusted deviations.	Middle-class	-0.2	2.3		-0.7
ze from a km	ce from a km km heer of physicians per province is not weighted. cates a sizable discrepancy between the adjusted and non-adjusted deviations. as are in bold.	Underprivileged	-1.7	-4.4		5.1
km km	km km km nber of physicians per province is not weighted. cates a sizable discrepancy between the adjusted and non-adjusted deviations.	Mixed	7.9			1
km km	km her of physicians per province is not weighted. cates a sizable discrepancy between the adjusted and non-adjusted deviations. as are in bold.	istance from a				
	of physicians per province is not weighted. s a sizable discrepancy between the adjusted and non-adjusted deviations. e in bold.	entre			0.20	
	of physicians per province is not weighted. s a sizable discrepancy between the adjusted and non-adjusted deviations. e in bold.	<100 km			2.1	
	Number of physicians per province is not weighted. Indicates a sizable discrepancy between the adjusted and non-adjusted deviations. Betas are in bold.	>100 km			-3.6	

(d' = -2.0) stand out as the provinces least favourable to selective abortion. Compared with the deviations of other provinces, Saskatchewan's position is very striking. The Atlantic provinces, Ontario, and Alberta were characterized by their midway stance. Other features that set Alberta apart were the large proportion of the variance accounted for by sociocultural factors, the number of significant factors, and the magnitude of observable disparities. These interprovincial differences are examined in greater detail in Chapter 8.

Religion was also a determining factor in physicians' attitudes toward selective abortion. It had a significant impact in six out of seven provinces. In each case, Catholics were far less favourable toward abortion. Conversely, Jewish respondents were much more favourable. This was reflected in adjusted deviations of 5 to 6 points above the mean for every province where there was a high number of Jewish physicians. The disparity between Catholic and Jewish respondents was particularly striking in British Columbia, with Catholics 10.2 points below the mean and Jews 12.7 points above it. Respondents who claimed no religious affiliation had attitudes that more or less approximated the stance of Jewish physicians toward abortion. This was also true of Anglicans and other Protestants (other than members of the United Church), but with a wider variation between provinces. The disparity between Jewish and Catholic respondents echoes what we had observed earlier regarding the perception of seriousness. Jewish physicians perceived anomalies as more serious and were more favourable to abortion, while Catholics, less concerned about anomalies, were also less favourable to abortion.

The influence of medical specialty was also clearly and consistently felt. Wherever this factor operated significantly — for Canadian physicians as a group and for Quebec, Ontario, and British Columbia physicians — GPs stood apart from radiologists and obstetricians in their attitudes toward abortion. General practitioners³³ were most opposed to abortion, while obstetricians and radiologists were most favourable to it. Paediatricians stood halfway between these two tendencies. This reflects what was observed in Chapter 5 through the bivariate analyses. The multivariate analysis has now confirmed that this difference between specialties does not depend on other factors related to a given medical specialty. There also appears to be a positive linear relationship between a medical specialty's use of technology and acceptance of abortion. Obstetricians and radiologists, who use the various procedures most frequently, were also the most favourable to abortion; paediatricians were next; and GPs, who have the least occasion to use PND technology and who were also the least favourable to selective abortion, were last.

Ethnic origin also had a significant influence on physicians' attitudes toward the acceptability of abortion.³⁴ Most notably, physicians of French origin were more favourable to selective abortion than those of other ethnic backgrounds. Conversely, Western European physicians (excluding the French and the British) were the least favourable. When other factors are

controlled for, those of French origin who practise outside Quebec were also more favourable to abortion than other respondents (n = 99, d' = +4.9). On the other hand, analysis of Quebec respondents' answers revealed that, within Quebec, physicians of French origin were least favourable to abortion while those of British (n = 41, d' = 4.7) or Jewish (n = 30, d' = 6) backgrounds were much more favourable. These Quebec-specific disparities are discussed in greater detail in Chapter 8.

Degree of religious practice was the only factor that operated significantly and consistently in every province. Across Canada, physicians who practised their religion were less favourable to selective abortion, and those who did not practise, or practised only occasionally, were more favourable. The differences were substantial, ranging from 6 to 12 points on a 12- to 60-point scale. The mean value of the scores on the "acceptability of abortion" scale also confirms this tendency of practising respondents to be less favourable to abortion than their non-practising colleagues, whatever their religious affiliation. The trend was nevertheless very pronounced among Catholics and Protestants. Churchgoers of these denominations were much less favourable to abortion than nonchurchgoers, and less than practising respondents of other religions. The proportion of non-practising Jews was rather small, and the gap between practising and non-practising Jews was, at any rate, very narrow. These findings were similar to those of other studies (see Chapter 1). Whereas the attitude toward abortion held by physicians belonging to a particular religion differed markedly from one province to the next (as was the case for Anglicans), the attitude of practising respondents was very consistent (whatever their religion). Therefore, religious practice was a more consistent predictor of physicians' attitudes toward abortion than was the actual religious affiliation.

Although statistically significant, a physician's age and number of children were the two weakest predictors of acceptance of abortion. As with the predictors discussed thus far, they were not consistent from province to province. Age was significant only in Alberta. Young Albertan physicians were the least favourable to abortion, older physicians the most. The influence of number of children was significant only in Quebec and in Alberta, where the trend was in the same direction; respondents with no children or only one were more favourable to abortion and those with several were less favourable. This finding was consistent with other studies.

The socioeconomic background of a physician's clientele had a significant impact on physicians' attitudes toward abortion in three provinces: Quebec, Saskatchewan, and British Columbia. Adjusted deviations, however, revealed no particular tendency. Alberta respondents close to a genetics centre were more favourable to abortion than those further away.

Analysis of "Directiveness on Abortion Decision"

Score distribution on the "directiveness on abortion decision" scale is presented in Figure 7.3. Scores ranged from 3 to 15 with a mean value of 8.8 and a standard deviation of 3. If the mean were reset on a 1 to 5 scale, it would be 2.9, which means that respondents as a group had an intermediate position on directiveness in abortion matters. Score distribution shows, however, that there was a fairly broad range of opinion among physicians on this issue. 35

The results of variance analysis on the "directiveness" scale are presented in Table 7.5. In addition to the acceptability of abortion, which strongly correlated with directiveness in abortion matters (r=0.42) and was therefore treated as a co-factor in analyzing the variance, eight factors showed a significant effect on directiveness. Together with "acceptability of abortion," these factors accounted for 24% of the total variance in directiveness scores, 10% being attributable to acceptability of abortion and 14% to sociocultural factors. These factors were province, religion, medical specialty, gender, practice area (urban or rural), and religious practice.

The MCA results are presented in Table 7.6, together with an analysis of results by province. The adjusted deviations show that the effects were rather minor. However, certain trends are indicated that warrant further investigation.

As was the case with the preceding analysis on abortion acceptability, there was a tendency among Catholic physicians to be more directive in abortion matters and, conversely, a tendency among Jewish and nonreligious physicians to be less so. Obviously, the more favourable physicians are to abortion, the less directive they will be in restricting women's access to it. There is also a tendency among female and urban physicians to be less directive. The gender-linked tendency was apparent in Quebec, Alberta, and British Columbia, whereas the tendency linked to area of practice was noted in Ontario and Manitoba. "Province" was also significantly associated with directiveness. Here again, three groups emerged: first, Manitoba and Alberta, with adjusted deviations of 0.31, were the most directive; second, Ontario (d' = -0.26) appeared to be the least directive; and third, the other provinces, the Maritimes, Quebec. Saskatchewan, and British Columbia, occupied a midway position on directiveness. This ordering of the provinces did not coincide with that observed for acceptability of abortion. Quebec, the most pro-abortion province, was not the least directive, nor was Saskatchewan the most directive.

This inverse relationship between the acceptability of abortion and directiveness was also not found for medical specialties, although a consistent trend appeared in the four provinces where this factor was significant. GPs, who were least favourable to abortion, were not the most directive. Indeed, they were slightly less directive than their colleagues. Paediatricians were the most directive, that is, the most inclined to restrict

Figure 7.3. Score Distribution on "Directiveness" Scale

Frequencies	
0	1.50 □
0	2.25 □
168	3.00 ***********
141	3.75 **********
4	4.50 I
207	5.25 **************
208	6.00 *************
340	6.75 **********************
2	7.50
337	8.25 ************************
329	9.00 *********************
308	9.75 ********************
3	10.50
431	11.25 ********************************
249	12.00 **********************************
202	12.75 ***************
1	13.50 l ·
62	14.25 *****
67	15.00 I *****
0	15.75 🗆
0	16.50 Ⅰ□
	+++++++++ 0 100 200 300 400 500
Descriptive	statistics 3 059 Standard deviation 3.017 Maximum 15
n Mean	8.752 Minimum 3 Missing values 13

access to abortion, followed closely by radiologists. In addition to referring to the "acceptability of abortion," directiveness includes the doctor/patient relationship. This might explain the disparities observed between physicians of different specialties. Since their concept of the role they play in their relationships with patients varies (see Chapter 5), they react differently to the idea of being directive. This stance can be fairly independent of their acceptance of abortion. Thus, although GPs were the least favourable to abortion, their relationship with their patients was apparently sufficiently liberal for them not to be the most directive. Paediatricians, on the other hand, assumed greater responsibility in their patients' decision-making process, even though their position on selective abortion was only moderate.

Lastly, religious practice appeared to be weakly associated with directiveness, with regularly practising respondents being more directive, occasional churchgoers somewhat less so, and non-practising physicians even less so.

Table 7.5. "Directiveness" Scale Variance Analysis

Source of variance	Sum of squares	DF	Mean squares	F	Sig of E
- Variation	- Squares		squares		Sig. of F
Covariates Acceptability of	4 520.027	1	4 520.027	648.280	0.000
abortion	4 520.027	1	4 520.027	648.280	0.000
Main effects	1 452.054	20	72.603	10.413	0.000
Religion	431.918	7	61.703	8.850	0.000
Province	121.243	6	20.207	2.898	0.008
Specialty	382.716	3	127.572	18.297	0.000
Gender	148.594	1	148.594	21.312	0.000
Area	115.447	1	115.447	16.558	0.000
Religious practice	129.257	2	64.628	9.269	0.000
Accounted for	5 972.081	21	284.385	40.788	0.000
Residual	19 331.557	2 773	6.972		
Total	25 303.638	2 794	9.056		

n = 3.072.

Number of missing values: 277 cases (9%).

Table 7.6. Adjusted Deviations for Each Predictor Having a Significant Bearing on Directiveness with Regard

to Abortion, for Car	Canada as a Whole and Each Province	hole and Ea	ch Province					
	Canada	Atlantic	Quebec	Ontario	Manitoba	Sask.	Alberta	BC/NWT
_	3 072	1	738	1 249		88	253	353
Z C	8.75		8.75	8.47		9.77	9.16	8.52
Standard deviations	3.02		3.02	2.91		2.83	3.00	3.06
R ²	0.24		0.24	0.21		0.26	0.39	0.41
R ² (factors)	0.14		0.15	0.14		0.09	0.22	0.30
R ² (abortion)	0.10	0.08	60.0	0.08	0.11	0.17	0.16	0.11
Drovince	0.07	-0.04	-0.02	-0.26	0.31	0.11	0.31	0.18
Pelinion	0.15		0.14	0.21			0.27	0.20
Catholic	0.40		0.2	0.8			-0.4	6.0
Anglican	-0.1		1	0.1			0.2	-0.4
United Church	-0-		ı	-0.7			-0.3	-0.1
Protestant	4.0		6.0-	0.4			1.6	9.0
Jewish	6.0-		-1.0	7.0-			-0.7	-0.6*
Oriental	0.2		,	6.0			-0.2	1
None	-0.5		-0.4	-0.4				9.0-
Capaialtu	0 13		0 14	0.13			0.16	0.15
GP	0 0		-0.4	-0.2			-0.1	-0.1
Ohstetrician	10-		-0.1	-0.2			-1.2	-0.2
Paediatrician	0.8		0.6	9.0			6.0	1.6
Radiologist	0.5		0.4	0.5			0.5	0.0
Gender	0.08		0.10				0.16	0.12
Male	0.1		0.2				0.3	0.2
Female	4.0-		0.0				ò	3

	Canada	Atlantic	Quebec	Ontario	Manitoba	Sask.	Alberta	BC/NWT
د	3 072	240	738	1 249	152	88	253	
Mean	8.75		8.75	8.47	9.40	9.77	9.16	
Standard deviations	3.02		3.02	2.91	2.80	2.83	3.00	
	0.24		0.24	0.21	0.19	0.26	0.39	
R ² (factors)	0.14		0.15	0.14	0.08	60.0	0.22	
R² (abortion)	0.10	- 1	0.09	0.08	0.11	0.17	0.16	0.11
Area Urban Rural	0.07 -0.1 0.4			0.09 -0.2 0.4	0.21 -0.4 0.8			
Religious practice Yes Occasional No	0.08 -0.3 -0.1		0.12 0.0 0.0 -0.4					
Ethnic origin British French Asian Jewish European East European		0.26 -0.2 -0.1 					0.19 0.3 -1.0 -0.6 - -0.83*	0.16 0.2 0.2 0.9
Distance from centre <100 km >100 km						0.27 0.7 -0.7		0.18 -0.4 0.7
Age Under 35 years 35 to 39 years 40 to 49 years			-0.4 -0.2 -0.2					

		0.2 E.1-	0.16 -0.9 0.0 0.4	
				tions.
.0.5 0.6	0.10 -0.9 0.2 0.1			ot weighted. n adjusted and non-adjusted devie
50 to 59 years 60 years or +	Number of children None 1 child 2 children 3 children or +	Clientele Middle-class Underprivileged	Physician's directiveness Low Medium High	Number of physicians per province is not weighted. Indicates a sizable discrepancy between adjusted and non-adjusted deviations. Betas are in bold.

Attitudes Toward Use and Development of Procedures

Ultrasound Scanning

As noted in Chapters 4 and 5, the number of ultrasound scans considered appropriate during a normal pregnancy, as well as the reasons considered valid for conducting the procedure, varied by province and medical specialty. The position of Quebec physicians on ultrasound scanning was diametrically opposed to that of Alberta and Manitoba respondents: the former use it more often, have more faith in it, and find greater justification for using it. More Saskatchewan physicians than the Canadian average also preferred two ultrasounds per pregnancy, but they were more reluctant to use ultrasound to screen for malformations and were less confident of its reliability. It should be emphasized that medical specialists were more prone to use ultrasound scanning and that physicians over 50 years of age were the most likely to use ultrasound scanning during a normal pregnancy. More than one-quarter of such respondents considered that at least two ultrasounds were needed. compared to 10% of those under 39 years of age. This trend was found in Quebec, Ontario, and Alberta. Practice area (urban, rural) and distance from a genetics centre also had an influence on the propensity to use ultrasound, but the effect was weak and rather inconsistent.

Expanded Access to Amniocentesis and CVS³⁶

Figure 7.4 shows the distribution on the "expanded access to PND" scale. The scale ranged from 3 to 48 with a mean value of 21.2. Central tendency measurements confirm that the distribution tends toward normal. As Figure 7.4 shows, Canadian physicians are somewhat inclined to restrict access to amniocentesis and CVS according to this scale.

As mentioned above, the analysis model was based on the assumption that the degree to which physicians consider anomalous conditions serious, the extent to which they find abortion acceptable, and their directiveness with regard to abortion had an influence on the likelihood they would increase or restrict access to amniocentesis and CVS. The following analyses include these three scales as co-factors, and their influence is estimated concurrently with that of sociocultural and professional factors. Overall, 15% of the variance is thus accounted for. The results of variance analysis are presented in Table 7.7.

Perception of seriousness (r = 0.19) and acceptability of abortion (r = 0.28) correlated positively with increased access to PND techniques. The more serious physicians considered anomalies to be and the more they accepted abortion, the more they were inclined to expand access to PND procedures. A physician's directiveness correlated negatively (r = -0.29) with increased access to the procedures. A "directive" physician was less likely to favour expanded access to the procedures. Directiveness had the greatest influence on the "expanded access to PND" scale, accounting for 3.8% of the variance, while "acceptability of abortion" and "perception of seriousness" accounted for only 1.2% and 0.9% respectively.

Figure 7.4. Score Distribution on "Expanded Access to Amniocentesis and CVS" Scale

requencies	Mid-point .
0	0.5 1 🗆
37	3.0 *
56	5.5 ***□
94	8.0 ****
161	10.5 ********
259	13.0 ***********
277	15.5 ************* [□]
315	18.0 ***************
601	20.5 ***********************************
289	23.0 *************
256	25.5 *************
261	28.0 ************
133	30.5 ******* □
112	33.0 ******□
76	35.5 ****
57	38.0 ***
40	40.5 I**
16	43.0
14	45.5 I*
9	48.0 I*
0	50.5 I
	++++++++++ 0 160 320 480 640 800
Descriptive	statistics 3 072 Standard deviation 8.1 Maximum 48 21.2 Minimum 3.4 Missing values 17

Table 7.7. Variance Analysis on Expanded Access to Amniocentesis and CVS Scale

Source of variance	Sum of squares	DF	Mean squares	F	Sig. of F
Covariates	21 927.829	3	7 309.276	132.230	0.000
Abortion	3 485.745	1	3 485.745	63.059	0.000
Gravity	1 480.456	1	1 480.456	26.782	0.000
Directiveness	5 953.124	1	5 953.124	107.696	0.000
Main effects	7 022.990	24	292.625	5.294	0.000
Medical specialty	2 693.690	3	897.897	16.244	0.000
Ethnic group	1 311.211	5	262.242	4.744	0.000
Religion	1 128.642	7	161.235	2.917	0.005
Area	806.927	6	134.488	2.433	0.024
Type of practice	720.043	2	360.022	6.513	0.002
Setting	487.739	1	487.739	8.824	0.003
Accounted for	28 950.819	27	1 072.253	19.398	0.000
Residual	150 351.271	2 720	55.277		
Total	179 302.091	2 747	65.272		

n = 3072.

Number of missing values: 324 cases (10.5%).

Sociocultural and professional factors accounted for 5% of the variance. In order of importance, the most significant were medical specialty, ethnic origin, religion, province of practice, type of practice, and practice area.

Professional characteristics, particularly medical specialty, had a significant and consistent impact (Table 7.8). GPs tended to be the least in favour of expanding access to PND; paediatricians and radiologists tended to be the most in favour. This trend existed in almost all regions of Canada. Obstetricians were more variable, but tended toward the position of members of the other two specialties.

Sociocultural variables had less impact. The influence of ethnic origin was considerable (beta = 0.10) but must be analyzed with caution to reflect its interactions with other variables such as "province" (for Quebec) or "religion" (for Jews). Generally, Jewish respondents were more favourable to expanded access and those of Asian background less favourable. In Manitoba and Saskatchewan, physicians of European (other than French or British) origin were noticeable by their less favourable attitude toward increased access.

Age had a substantial impact in the four western provinces, but no consistent trend was discernible. In Saskatchewan, age accounted for 21%

Table 7.8. Adjusted Deviations for Each Predictor Having a Significant Bearing on Expanded Access to Amniocentesis and CVS, for Canada as a Whole and Each Province

	Canada	Atlantic	Quebec	Ontario	Manitoba	Sask.	Alberta	BC/NWT
	3 072		738	1 249	152	88	253	
Mean	21.2		22.2	21.4		20.7	20.8	
Standard deviations	8.1		8.5	7.8		9.1	8.23	
R^2	0.15		0.14	0.12		0.51	0.43	
R ² (factors)	0.05		0.04	0.03		0.23	0.24	
R ² (co-factors)	0.1	0.08	0.1	60.0		0.28	0.19	0.15
Specialty	0.11	0.17	0.18	0.11			0.16	0.18
GP	-0.58	-0.69	-1.77	-0.45			-0.44	-0.65
Obstetrician	0.06	3.87	0.61	-0.74			-2.09	2.32
Paediatrician	1.73	0.25	1.9	1.6			2.19	4.31
Radiologist	1.5	1.68	9.0	0.92			3.27	0.45
Ethnic origin	0.00				0.30	0.31		
British	-0.27				2.52	0.97		
French	0.46				-2.24	-1.70		
Asian	-1.61					-2.58		
Jewish	-0.82*							
European	0.33				-2.32	-3.52		
East European	1.72				-0.21	4.07		
Medical school							9	7
attended		0.18			0.29		0.18	0.15
Newformalland		4 22						
Own province					1.52		1.30	1.46

	Canada	Atlantic	Quebec	Ontario	Manitoba	Sask.	Alberta	BC/NWT
c	3 072		738	1 249		88	253	
Mean	21.2		22.2	21.4		20.7	20.8	
Standard deviations	8.1		8.5	7.8		9.1	8.23	
. T2	0.15		0.14	0.12		0.51	0.43	
R ² (factors)	0.05		0.04	0.03		0.23	0.24	
R ² (co-factors)	0.1	0.08	0.1	60.0	60.0	0.28	0.19	0.15
Other Canadian Great Britain		-1.18			0.26		-0.71	-0.71
Other		1.10			-3.78		-2.36	0.73
Age					0.33	0.54	0.17	0.16
Under 35 years					3.03	4.61	96.0-	-2.07
35 to 39 years					-1.27	2.98	1.55	0.28
40 to 49 years					-2.43	-3.63	-0.82	1.14
50 to 59 years					1.09	7.10	-1.86	-1.35
60 years or +					4.19	-5.37	3.11	-2.48
Early convert (to								
technology)			0.21		0.08			0.16
No		-1.69		-0.77				-1.76
Average		-0.01		0.24				1.32
Yes		2.61		0.71				0.18
Number of children		0.29					0.20	0.16
None		5.63					2.05	-2.20
1 child		1.38					-0.55	-3.80
2 children		-1.52					1.40	0.18
3 children or +		-0 95					1 10	

	0.23 -1.68 3.99 1.9 -1.89 -2.56 0.02	
0.19 -0.63 -0.86 3.06		0.2 -1.03 2.34
		ijusted deviations.
0.2 -1.7 2.16 0.51		ans per province is not weighted. discrepancy between adjusted and non-adjusted deviations.
	0.09 -0.69 0.78 1 0.47 -0.32 0.05 0.05	ns per province
Religious practice Yes Occasional No	Religion Catholic Anglican United Church Protestant None Jewish Oriental Other Area Urban	Obstance from centre <100 km >100 km 1 Number of physicians per province is not weighted. * Indicates a sizable discrepancy between adjusted a Betas are in bold.

of the variance in the "expanded access to abortion" scale; in Manitoba, 10%.

Predisposition Testing

We asked physicians to tell us when (*in utero*, at birth, in adulthood, or never) and why (preventing births, early treatment, preventive counselling, or no possible reason) they would use genetic predisposition testing if it became available. Both questions were asked for five diseases (diabetes, alcoholism, schizophrenia, Alzheimer's disease, and coronary heart disease).

Generally, respondents were lukewarm to the possibility of using the tests *in utero*. The cross-tabulations for each of the diseases, times, and reasons show that respondents were remarkably consistent. Those who believed in preventing the birth of afflicted children would conduct the tests prenatally, those whose aim was early treatment or preventive counselling would use them at birth or in adulthood, and those who believed that no possible reason could justify testing for predispositions would never use them.

For all the conditions listed the oldest physicians were the most favourable to using prenatal testing, with *in utero* diagnosis to prevent the birth of afflicted children. This trend is apparent in Figure 7.5, along with that of rejection of predisposition tests.

The other influential factor was specialty. For three disorders (diabetes, schizophrenia, and Alzheimer's disease), GPs were less favourable³⁷ to the use of predisposition testing. Conversely, obstetricians were more inclined to use it.

It would therefore appear that GPs and younger physicians are the most reluctant to use predisposition testing to prevent the birth of children who may develop diseases later in life.

Funding Priorities

In question 13, physicians were asked to set health budget allocation priorities. They had to rank seven items, ³⁸ which we grouped together in two main classes: funding for medical technology (items 2, 3, 4, and 7) and funding for preventive medicine (items 1, 5, and 6).

The findings show that physicians generally tended to favour spending for the prevention and treatment of pregnancy problems. Means and standard deviations for each of the seven items, in descending order, are given in Table 7.9.

The integrated nutritional assistance programs for women who are at risk of having babies with low birthweights received the most support from Canadian physicians. No less than 66.3% said these programs should be one of the top two spending priorities. Also well supported were information programs about the harmful effects of alcohol and smoking (58.6%), and multidisciplinary teams to provide care to socially disadvantaged pregnant women (49%). Conversely, only 6.2% of Canadian physicians felt that developing cytogenetic and obstetrical ultrasound

Table 7.9. Means and Standard Deviations of Funding Priorities, in Descending Order

	Mean	Standard deviation
Integrated nutritional assistance programs to reduce number of babies with low birthweights	2.41	0.03
Information programs on harmful effects of smoking and alcohol	2.50	0.03
Multidisciplinary teams for disadvantaged communities	2.71	0.03
Introduce population-wide blood tests	4.67	0.03
Improve ultrasonography training and increase the number of specialists	5.11	0.03
Increase and improve cytogenetic laboratories	5.17	0.03
Develop infertility treatment services	5.27	0.03

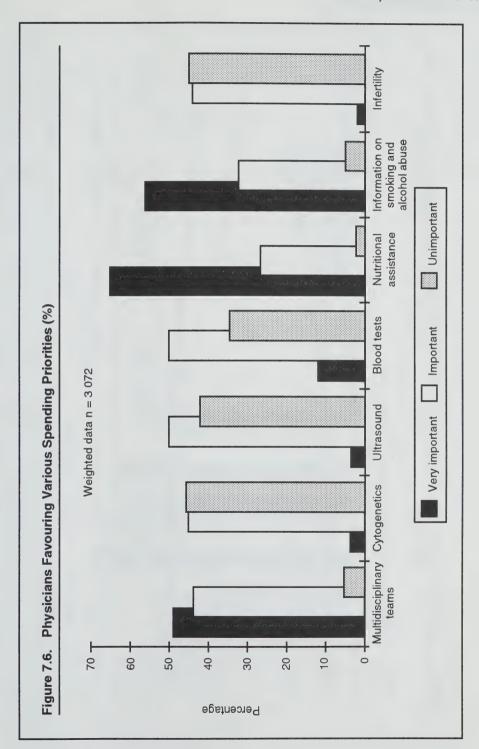
^{*} Note that items are ranked in descending order of importance. The lower the mean, the more important funding was considered (1 being the most important, 7 the least).

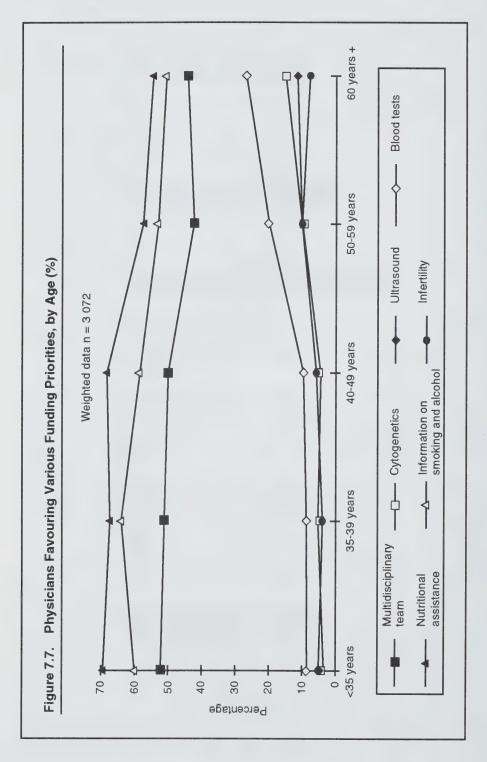
services should be one of the top two spending priorities, 12.9% gave priority to prenatal blood test screening programs, and 6% were in favour of services for the treatment of infertility. These differences are shown in Figure 7.6.

Multivariate analysis identified the factors that had a significant impact on these priorities. The six factors revealed are, in order of importance, age, specialty, distance from genetics centre, province, ethnic origin, and religion. The results of this analysis are set out in Table 7.10.

As Figure 7.7 shows, the older the physicians were, the more they tended to favour funding of preventive medicine and technology. Specialty also had an influence on priority setting (Figure 7.8). GPs were more likely than the other specialties to give priority to preventive action, mainly the formation of multidisciplinary teams and information campaigns.

While the impact of the other four factors (province, distance from genetics centre, ethnic origin, and religion) was statistically significant, there was no clear systematic tendency. Quebec physicians differed from their Canadian colleagues on the development of cytogenetics laboratories and public information campaigns: more of them thought that the development of cytogenetics laboratories was a major priority and that information campaigns were not. Tables 7.11, 7.12, and 7.13 show the importance accorded to the various funding items by physicians, broken down by each of these four factors and subsets.





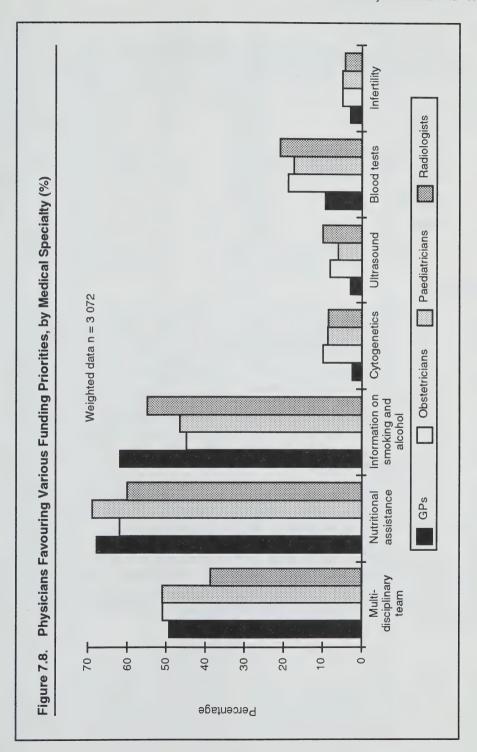


Table 7.10. Multivariate Analysis of Funding Priorities

	Wilks	F approx.	Sig. of F
Age	0.88	6.63	0.000
Specialty	0.91	5.69	0.000
Distance from genetics centre	0.95	5.60	0.000
Province	0.88	4.43	0.000
Ethnic group	0.93	2.67	0.000
Religion	0.91	2.34	0.000

Summary

Our analysis shows that there are major variations in physicians' perceptions of the seriousness of various anomalies, their acceptance of abortion, their directiveness with regard to abortion, and their tendency to want to expand or restrict access to PND techniques. Most of the variations in perception of seriousness remain unexplained in our analysis and must be attributed to individual differences. However, this perception appeared to be strongly associated with physicians' attitudes toward abortion. The more they perceived anomalies as serious, the more accepting they were of abortion. In addition, the variation in their acceptance of abortion may be explained in large part by sociocultural and professional factors. Physicians' religion, religious commitment, province of practice, medical specialty, place of study, proximity to a genetics centre, number of children, age, and gender all determine their attitudes toward abortion. These attitudes, in turn, determine the degree of directiveness with respect to the decision to terminate a pregnancy: the more accepting they are of abortion, the less they will tend to dictate behaviour to the parents. Several factors that influence the "perception of seriousness of anomalies" and "acceptability of abortion" also influence a physician's directiveness. Physicians' attitudes toward PND procedures are, of course, influenced by their attitudes toward anomalies and abortion, but age and specialty also play a role.

Table 7.12. Phys	Physicians with	icians with Various Funding Priorities, by Ethnic Background (%)	ng Priorities,	by Ethnic B	ackground ((%)	
	Multidisciplinary team	Cytogenetics laboratory	Ultrasound	Ultrasound Blood tests	Nutritional assistance	Nutritional Information on assistance smoking and alcohol Infertility	Infertility
British	51.7	4.0	3.9	10.4	67.8	61.1	5.2
French	54.7	6.6	9.7	13.9	64.8	46.4	8.6
Asian	41.8	7.1	7.2	21.3	63.8	60.4	6.8
Jewish	40.7	10.6	13.6	28.9	63.8	43.4	7.8
European	42.1	6.5	4.7	12.5	68.7	67.3	2.0
Eastern European	43.1	7.2	0.6	12.0	64.8	61.0	6.4

Infertility 8.3 6.9 7.1 12.0 on smoking and Information alcohol 60.2 52.3 63.5 61.5 63.1 53.6 54.1 Table 7.13. Physicians with Various Funding Priorities, by Religious Background (%) assistance Nutritional 62.6 65.3 68.4 66.9 64.7 59.3 **Blood tests** 13.2 14.0 10.8 11.2 19.3 20.4 Ultrasound 7.6 5.7 4.8 2.8 7.8 13.8 Cytogenetics laboratory 10.8 8.3 7.1 6.2 6.4 Multidisciplinary 43.2 46.6 48.7 46.5 47.1 United Church Protestant Anglican Catholic Oriental Jewish None

Chapter 8. Discussion of Interprovincial Differences

In this chapter, we present an overview and discussion of our major findings concerning the influence of sociocultural and professional factors on physicians' attitudes and opinions regarding PND. Particular emphasis is placed on differences observed between Canadian provinces, because interprovincial cleavages have proved to be the most consistent and the most substantial.

Summary of Multivariate Analyses

First, we shall describe the profile of Canadian physicians with respect to PND. If we reset all scales to their original 1 to 5 spread, with 1 meaning extremely unfavourable and 5 extremely favourable, we see that Canadian physicians take a middle-of-the-road position on PND. Canadian physician perceives anomalies overall as fairly serious (mean = 3.16), is moderately directive (2.92), and is somewhat opposed to abortion (2.62) for the conditions specified, which cover a broad spectrum. The same attitude was observed with regard to the use of technology, since the status quo is generally accepted. This was particularly true with regard to age 35 as the eligibility threshold for amniocentesis, and the use of one ultrasound scan in the course of a normal pregnancy. As far as developing PND procedures is concerned, however, Canadian physicians are reticent. They are slightly opposed to expanded access to amniocentesis and CVS. Predisposition testing is considered valuable if used at birth or in adulthood. Lastly, Canadian physicians would give higher priority to funding prevention-oriented social programs than medical technology.

There were, however, broad differences in physicians' attitudes. Table 8.1 presents the main results of the mutivariate analyses. The direction of the most substantial³⁹ cleavages is indicated by + or -. Province and medical specialty were the only factors to have a decisive influence on *each* attitude and opinion regarding PND, both those of an ethical or moral nature (perception of seriousness of anomalies, acceptability of selective abortion, and directiveness) and those of a professional or medical nature (expanded access to amniocentesis and CVS, use of obstetrical ultrasound scanning, use of genetic predisposition testing, and funding priorities). The other factors had an influence only on issues of an ethical or moral nature: religious affiliation, degree of religious practice, ethnic origin, number of children, gender, and practice area influenced physicians' attitudes regarding the acceptability of abortion, directiveness in abortion matters, and the seriousness of anomalies.

The reasons for this are evident. When physicians must form an opinion on a medical matter, their reference is their professional environment, which includes the norms for their type of practice and the opinion of their fellow specialists. On the other hand, medicine is often at a loss when it comes to moral or ethical issues. To form an opinion,

			,		Procedures	ures	
	Seriousness	Abortion	Directiveness	Amnio./CVS	Ultrasound	Predisp. test	Priorities
Provinces							
Atlantic						ı	1
Quebec	+	+			+		+
Ontario			ı			ı	
Manitoba		i	+		1		
Saskatchewan		ı			+	+	
Alberta			+		ı		
BC/NWT		+				+	
Religion							
Catholic	1	ı	+				
Protestant	i						
Jewish	+	‡	1				
None		+	8				
Anglican	+						
United Church Other		+					
Specialty							
GP		i	ı	ı	ı		I
Paediatrician	ı		+	+	+		+
Obstetrician		+	ı	+	+	+	+
Radiologist		+	+	+	+		+
Ethnic origin							
English Can.	8						
French Can.	+	+					
European		1					

	+	•		+				+		+		1	+	
	+	1		1	+		+	ı	ı	+				
Number of children	0 or 1	3 or +	Gender	Male	Female	Setting	Urban	Rural	Religious practice	None or occasional	Age	<40 years	>50 years	

physicians, like anyone else, must rely on their personal vision of the world, its standards, and its values. This is where sociocultural factors (religion,

religious practice, etc.) can have an important influence.

Physicians' attitudes are strongly interdependent. We observed that attitudes appeared to influence each other in the following sequence: perception of seriousness of anomalies \rightarrow acceptability of abortion \rightarrow directiveness \rightarrow attitude toward PND techniques. As a result, the more physicians considered anomalies to be serious and the more accepting they were of selective abortion, the less directive they were (in the sense of restricting access to abortion) and the more they favoured the use and development of PND procedures.

The greatest influence was undoubtedly that of religion. A Catholic physician sees anomalies as less serious, is much less accepting of abortion, and is more inclined to restrict access to it. A Jewish physician's attitudes are at the other end of the spectrum. Physicians without religious affiliation are, like Jewish physicians, more accepting of abortion and less directive. Religious practice also has a determining influence on the acceptability of abortion. Whatever their faith, physicians who practise their religion are less accepting of abortion than those who practise only occasionally or not at all.

The influence of professional factors is mainly observed in the divergent attitudes and opinions of GPs and specialists. GPs are less accepting of abortion and the use or development of technology, yet they are the least directive. Conversely, radiologists and obstetricians, who are the most in favour of abortion, are paradoxically the most directive. Different concepts of the physician-patient relationship are probably involved.

Canadian physicians of French origin consider anomalies more serious and are more in favour of abortion. Those of European origin (other than British or French) are less in favour. Physicians with few children are also more inclined to view anomalies as serious and to accept selective abortion. Female physicians and those who practise in an urban setting see anomalies as more serious and are less directive in abortion matters.

Compared with their younger colleagues, older physicians are more in favour of using PND technology, more ultrasound scans during pregnancy, and earlier use of genetic predisposition testing; they are slightly more favourable to funding medical technology.

Interprovincial Differences

Throughout this report, we have observed the crucial role played by the provinces in shaping physicians' attitudes and opinions toward PND technology and the ethical dilemmas it creates. The sociocultural and professional profile of a province's physicians certainly explains in part the attitude of the physicians practising there. One can understand, for instance, that selective abortion is more accepted in a province with a large

percentage of obstetricians and physicians who do not practise their religion. However, as noted in Chapter 7, these sociocultural characteristics alone cannot explain the observed differences between provinces.

Provinces are, in fact, regions where a host of factors may shape physicians' opinions and attitudes. These may include, for example, the differences in the organization of health care services, the availability of PND services, public opinion, women's groups, associations for the disabled, lawsuits against certain physicians, and so on. In a way, each province has its own "culture," in the sociological sense of the term, above and beyond the characteristics of its individual citizens. Obviously, we could not examine all of those provincial particularities within the framework of the present study. We were able, however, to evaluate the influence of province of residence statistically by controlling for physicians' sociocultural and professional characteristics as measured by the questionnaire.

We conducted a cluster analysis⁴⁰ to identify provinces with the most similar attitude profiles. Similarities between provinces were based on provincial means for the various scales (i.e., the "perception of seriousness." 'acceptability of abortion," and "directiveness" scales), plus four "use or development of procedures" measurements: (1) expanding the criteria for access to amniocentesis and CVS scale; (2) number of ultrasound scans considered appropriate during a normal pregnancy; (3) early use of predisposition tests;⁴¹ and (4) preference given to technology funding.⁴² In order to give equal weight to each characteristic on which similarities between provinces were evaluated, all scales were standardized (converted into scales with a mean value of 0 and a standard deviation of 1).

Table 8.2 presents provincial means for the various standardized It shows, for example, that the standardized mean of the "acceptability of abortion" scale for Quebec physicians is 0.31 (compared to 35.3 on the original 12-60 scale) and -0.59 for Saskatchewan (23.7 on the original scale).

Four groups of provinces emerge from this analysis: (1) Ontario and British Columbia, (2) Manitoba, Alberta, and the Atlantic provinces. (3) Saskatchewan, and (4) Quebec. Quebec and Saskatchewan are very different from each other, as well as from other provinces.

To show these differences more clearly, standardized means of the "perceived seriousness," "abortion," and "directiveness" scales for the seven provinces are presented in Figure 8.1. Saskatchewan is clearly the province where physicians are most opposed to selective abortion and most directive. However, the province does not stand out with regard to "perception of seriousness of anomalies." At the opposite end, Quebec clearly stands out as the province most accepting of abortion and where anomalies are seen as most serious. The chart also shows the similarities between the other provinces. Manitoba, Alberta, and the Atlantic provinces share more or less the same attitude profile, slightly below the Canadian average with regard to "perception of anomalies" and "acceptability of

Table 8.2. Provincial Means on the Various Standardized Scales

	Serious- ness	Abortion	Direct.	Amnio.	Ultra- sound	Diag.	Priorities
Atlantic	-0.16	-0.14	-0.04	-0.21	-0.18	-0.16	-0.33
Quebec	0.48	0.31	0.00	0.11	0.06	0.12	0.29
Ontario	-0.11	-0.01	-0.09	0.01	-0.03	-0.06	0.08
Manitoba	-0.03	-0.21	0.22	0.08	-0.53	-0.16	-0.15
Saskatchewan	0.00	-0.59	0.34	-0:07	0.37	0.15	-0.15
Alberta British	-0.14	-0.14	0.14	-0.06	-0.44	0.02	-0.14
Columbia	-0.03	0.11	-0.08	0.02	-0.02	0.07	-0.04

abortion," and slightly above for directiveness. Ontario and British Columbia are very close to the Canadian average.

Figure 8.2 gives the adjusted means for the four "use of procedures" measurements for each province. Except for Saskatchewan, the attitude profiles of the various provinces were very similar to those for acceptability of abortion. The attitudes of Ontario and British Columbia physicians closely match the Canadian average. Physicians from the Atlantic provinces, Manitoba, and Alberta present more unfavourable attitude profiles to PND techniques. They would order fewer ultrasound scans in the course of a normal pregnancy, would use predisposition tests at a later stage, would allocate more funds to preventive health programs, and (except for Manitoba physicians) would be less inclined to expand access to amniocentesis. Physicians in Saskatchewan, and especially those in Quebec, were more sympathetic to PND techniques. Quebec physicians would be more sympathetic to expanding access to amniocentesis, order more ultrasound scans in the course of a normal pregnancy, perform predisposition tests at an earlier stage, and favour funding technology more than other physicians. Saskatchewan physicians would be more in favour of using ultrasound scanning during pregnancy and also predisposition tests.

Figure 8.3 provides an overview of observed variations between provinces, presenting the standardized means for each province in graph form. Once again, we see how different Quebec and Saskatchewan are relative to each other and relative to the other provinces. One also observes how similar the profiles of Manitoba, Alberta, and the Atlantic provinces are on the one hand, and those of Ontario and British Columbia on the other. The latter provinces have no distinctive features as their physicians' attitudes essentially reflect the Canadian average.

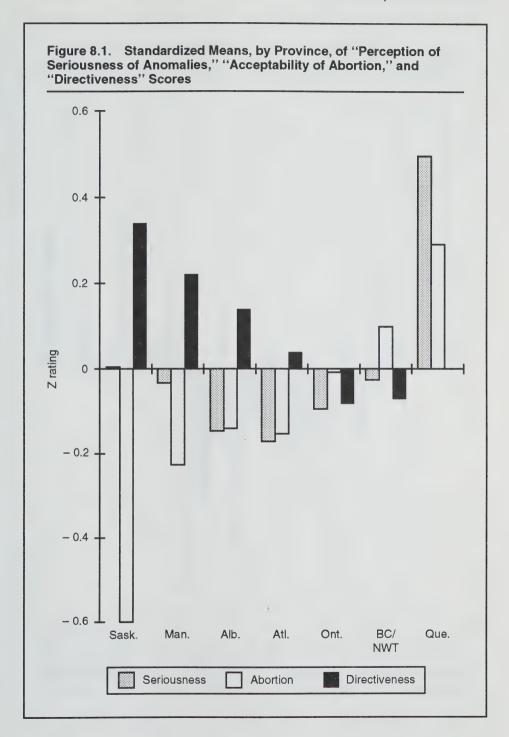


Figure 8.2. Standardized Means, by Province, of Opinion on Use of PND Procedures Scales 0.6 0.4 0.2 Z rating 0 - 0.2 - 0.4 - 0.6 Sask. Man. Alb. Atl. Ont. BC/ Que. **NWT** Predisposition Medical technology Amniocentesis Ultrasound tests funding

Figure 8.3. Standardized Means, by Province, of Attitude and Opinion on Selective Abortion and PND Procedures Scales 0.6 0.4 0.2 0 - 0.2 - 0.4 Sask. Man. Alb. Atl. Ont. BC/ Que. - 0.6 NWT Amniocentesis Abortion Directiveness Seriousness Predisposition Ultrasound Medical technology funding tests

What Is Going on in Quebec and Saskatchewan?

Whatever the analytical tool employed or the measurement considered, Saskatchewan and Quebec were always at opposite poles, and relatively far removed from the Canadian averages, which were better reflected in Ontario and British Columbia.

As multivariate analysis progressed, we also observed that a large number of cultural and professional factors interacted with "province" to explain the attitude variations. Although each of these factors had a low impact when taken individually, collectively they explained from 5% to 30% of the variance depending on the attitude examined, which is considerable in social science studies. It should be noted, however, that even when these factors were statistically controlled for, province of residence continued to have an independent influence, which suggested that, all other things being equal, "distinct" provincial cultures do exist.

We shall now examine Quebec and Saskatchewan more closely. It should be recalled that 30% of Saskatchewan physicians said they were totally opposed to selective abortion, whereas this percentage was 5% in Quebec and 14% for Canada as a whole.

The particular combination of cultural and professional characteristics of these two provinces sheds some light on why they have such different attitudes. The main characteristics of each province are as follows:

- Quebec's ethnic composition is mostly French;
- with regard to PND, Quebec has the largest proportion of specialists in Canada;
- the obstetrical practice of Quebec GPs is more intensive and specialized than that of GPs in other provinces;
- Quebec has the largest proportion of Catholic physicians in Canada;
- Quebec physicians were the least likely to claim they practised their religion;
- of all Canadian physicians, those in Quebec are the most heavily concentrated in urban areas, judging by their answers; and
- they were the least likely to identify their clientele as underprivileged.

Saskatchewan's profile is at the opposite pole:

- the province has the largest proportion of GPs in Canada;
- it has the most Catholic physicians after Quebec;
- of all Canadian physicians, those in Saskatchewan are by far the most likely to practise their religion; it should be noted that 85% of the province's Catholics claim to practise their religion compared to 25% in Quebec;

- Saskatchewan physicians are the most likely to say that they practise in a rural setting;
- their clientele is apparently the most underprivileged in Canada;
- Saskatchewan physicians are on average the oldest in Canada and have a much larger number of children than average.

With such different profiles, it is not surprising that these two provinces are so far apart in terms of physicians' acceptance of selective abortion, their perception of seriousness of anomalies, and their directiveness, in the sense of restricting access to abortion. The fact that the Quebec medical profession is more specialized undoubtedly also accounts for the difference in views on amniocentesis and funding priorities.

However, as noted in Chapter 7, these characteristics alone cannot explain all the differences. For example, since Catholicism is associated everywhere in Canada with a lower acceptance of abortion, it could have been expected that the Catholicism of Quebec's Francophone physicians, even if somewhat tempered by a low degree of religious practice, would have made abortion less acceptable. This was not the case. It could also have been expected, since physicians of French origin in Canada are the most sympathetic to abortion, that Quebec's Francophone physicians would be more favourably disposed toward abortion than Quebec's Anglophones (of British or Jewish origin). This was not the case either.

In other words, where PND is concerned, there definitely appears to be a province-specific "culture" in Quebec, and no doubt in the other provinces as well (or at least in the three other groups identified above). Taken together, the sociocultural characteristics of a province form a whole that is greater than the sum of its parts. This whole, or culture, can have an independent, supplementary impact on the attitudes of a physician over and above the influence of each one of the characteristics.

Special Case of Quebec Anglophones

The distinguishing mark of Quebec's "culture" is that it has its roots in two large linguistic groups, the one Francophone and the other Anglophone. Based on our previous surveys, one question concerned us throughout our analysis: How do the attitudes of Quebec's Anglophone physicians compare with those of their colleagues in other provinces, and how close are they to those of their Quebec Francophone colleagues?

We compared the attitudes of Quebec's Anglophone physicians (practising mostly in Montreal) with those of physicians practising in a comparable environment (Toronto), those of Quebec's Francophone physicians, and then those of other Canadian physicians generally.

This analysis is summarized and presented in Table 8.3. The penultimate column shows the percentage of Canadian physicians favouring a particular attitude. The other columns give the deviation from the Canadian percentage for four groups of physicians; Anglophone Quebeckers, Francophone Quebeckers, Toronto physicians, and other Canadian medical practitioners. The symbol 1 in the last column indicates

Table 8.3. Main Differences Between Toronto Physicians, Quebec's Anglophone and Francophone Physicians, and Physicians in Rest of Canada

	Que. Franco.	Que. Anglo.	Toronto	Other	Canada %	Trend
Ultrasound scanning Favour						
- doing no ultrasound	-15.7	-13.8	3.2	2.7	19.6	
- doing 1 ultrasound	-5.4	-17.4	-8.3	1.9	63	1
doing 2 ultrasoundsusing ultrasound to	20.3	30.2	2.9	-4.4	16	†
screen for malformations - using ultrasound to	30.4	17.9	3.8	-5.6	61.1	1
reassure a woman - using ultrasound to conform to professional	4.1	10.7	10.3	-1.6	28.4	=
practice Do not favour using	-2.8	10	-1.6	0.1	15.2	
ultrasound to view fetus Do not favour using ultrasound to give women	-10.7	-5.8	3.1	1.7	77.1	
sense of responsibility Accept refusal of ultrasound because exam	-7.8	-0.8	8.5	0.8	65.1	
is not important Do not accept refusal and suggest to see another	-5.2	-14	0.8	1.3	26.5	
physician	7.2	2.9	-0.9	-1.2	2.8	
Reliability of ultrasound scanning Confident that ultrasound can diagnose						
- limb malformations	7.5	-3.5	-2.4	-0.9	41.9	1
- hydrocephaly	-1.4	14.4	6.3	-0.7	44.9	Ť
- spina bifida No confidence in ultrasound for trisomy 21 without structural	7.3	1.7	-0.4	-1.1	38.8	·
malformations	7.1	2.6	-6.2	-0.8	87.4	

Table 8.3. (cont'd)

	Que. Franco.	Que. Anglo.	Toronto	Other	Canada %	Trend
Eligibility criteria for amniocentesis Accept that it be available						
without criteria (public system) Accept that it be available	10.8	16.3	-1.7	-2.1	14.2	
without criteria (against payment) Accept 33-year-old	-13.2	-4.5	2.6	2.1	59	
woman's request if she assumes risks Anxiety is a valid reason	5.2	16.5	0.4	-1.5	21.9	
for using amniocentesis	8.3	10.6	-3.4	-1.5	21.7	
Expand eligibility to women under 35	-1.2	9.2	2.6	-0.3	9.7	1
Limit eligibility to women 40 and over	-2.7	-6.5	-3	0.9	7.2	\
Age of eligibility for CVS Under 34 years 35 years Never	-2 -12.4 11.6	9.9 3 0.3	-0.8 6.7 -6.8	-0.1 1.4 -1.3	8.3 51.4 13.5	
Directiveness with regard to amniocentesis Advise to go through with						
procedure (36-year-old woman) Advise to go through with	21.3	27.6	-1.5	-4.3	44.7	↑
procedure (38-year-old woman) Advise not to go through	21.5	19.5	-1.1	-4	62	↑
with procedure (36-year-old woman)	-5.7	-7.1	-6.8	1.6	15	
More frequent use of PND because of the fear of lawsuits	-17.2	-12	-4.6	3.4	55.8	

Table 8.3. (cont'd)

	Que. Franco.	Que.	Toronto	Other	Canada	
	Δ%	Aligio.	A%	Δ%	%	Trend
Predisposition tests						
Favour testing in utero for:						
- diabetes	2.9	7	4.8	-1	7.5	↑
- schizophrenia	9.6	10.3	7.1	-2.3	17.4	1
- Alzheimer's disease	5.1	7.4	7	-1.5	8.5	=
Favour testing to prevent						
births:						
- schizophrenia	10.5	9.9	3.8	-2.3	16.5	↑
- Alzheimer's disease	3.7	6.7	5.1	-1.1	7	\uparrow
Do not favour self-						
prescribed tests to identify						
fetal sex	9.9	-9.1	2.6	-1.3	69	
Methods for						
circumventing genetic disorders						
Approve of advising couple						
to use artificial insemination						
when the female partner						
has a genetic disorder	-11	3.8	0.0	. 4	74.0	
Approve of surrogate	-11	3.6	9.9	1	74.3	
motherhood	-9.6	10.4	0.4	0.5	40.4	
Mothernood	-9.0	10.4	9.4	0.5	40.4	=
Perception of						
seriousness						
Perceive as serious or						
difficult:						
- learning difficulties	11.9	6	4.3	-2.3	45.5	↑
- cleft lip and palate	13.7	4.4	10.7	-2.9	48.8	
- lobster claw deformity of						
the hand	17.3	16.8	8.3	26.2	41.1	↑
- intellectual impairment	26.6	9.8	6.2	-4.8	61.1	$\uparrow \uparrow $
- paraplegia	11.5	8.5	3.5	-2.4	83.7	†
- female sterility	6.1	12	7.9	-1.9	15.1	†
- male sterility	6.8	14.2	4.9	-1.9	13.1	1
- hypogonadism	6.4	16.5	6.6	-2.1	21.6	†
- XYY syndrome	0.5	16.4	5.9	-1.1	21.7	†
- XXY syndrome	3.1	18.2	3.9	-1.5	27.7	†
- XXX syndrome	4.5	10.8	3.4	-1.4	23.6	†
Would not accept the idea			0	-1,-	20.0	1
of having a child with						
trisomy 21	27.1	19.5	7.3	-5.4	40.2	1

Table 8.3. (cont'd)

	Que. Franco.	Que. Anglo.	Toronto	Other	Canada %	Trend
Acceptability of abortion Abortion acceptable for:						
 trisomy 21 without structural malformations Duchenne muscular 	18.2	23.9	14.8	-4.6	51.1	↑
dystrophy	2.8	25.5	16	-2.3	51.3	\uparrow
- Huntington's disease - severe heart	-0.2	27.5	11.3	-1.7	51.3	†
malformations	11.6	8.1	11.7	-2.8	43.2	
- cystic fibrosis	5	28.6	11.7	-2.6	37.4	1
spina bifida	6.2	18.5	16.8	-2.6	29.3	=
phenylketonuria	5.8	17.2	7.3	-1.9	21.6	=
Turner's syndrome	4	15.3	5.8	-1.5	21	1
Klinefelter's syndrome	-0.1	23.2	12.3	-1.5	17.2	1
XYY syndrome	-0.5	18.9	9	-1.2	16.3	1
XXX syndrome lobster claw deformity of	0.9	17.5	10.6	-1.4	15.9	1
the hand	2.2	15.5	9.3	-1.4	10.4	\uparrow
Aborting in the first trimester is more justified than in the second PND done with the deliberate intention to abort must be condemned	3.1	14.7	0.4	-1 -1.4	46.7 18	↓
Directiveness with regard o abortion						
The parents' freedom of choice with respect to abortion is an absolute right Opposed to physicians,	6.6	15.3	11.6	-2.3	49.6	↑
and not parents, deciding which anomalies warrant pregnancy termination	-16.8	4.5	8.2	1.9	59.5	
Information disclosure Disclose information on fetal sex only for medical						
reasons In some circumstances,	-10.8	-11.2	-7.4	2.5	36.5	\
feel legally bound to disclose information	4	10.5	3.6	-1.2	39.8	\uparrow

	Que. Franco.	Que. Anglo.	Toronto	Other	Canada %	Trend
Perception of the counterproductive effects of PND (eugenics) Believe that further advances in PND will make "disorders" out of conditions previously						
considered normal PND increases intolerance	-2.2	-11	-7.1	1.2	50.9	1
of abnormality Giving birth to a genetically abnormal child at a time when PND and abortion are available is socially	13.1	-5	-1	-1.8	49	
irresponsible Favour enacting laws to control the transmission of genes causing severe	10.5	13.4	4.3	-2.4	16.3	1
diseases	8.2	7.1	-1.5	-1.5	13.5	
Funding priorities Do not agree that PND cannot be a priority considering the number of						
problems of social origin	8.7	14	10	-2.4	35.5	

where Quebec Anglophone physicians have a more sympathetic attitude than Toronto physicians and where the latter have a more sympathetic attitude than other Canadian physicians. A \downarrow symbol indicates the opposite trend.

The frequent occurrence of these symbols clearly indicates a consistent trend in the answers of these three groups of physicians. The same trend appears for use of ultrasound scanning, amniocentesis, and predisposition tests, as well as a lower eligibility age for amniocentesis. Quebec's Anglophone physicians were most in favour of such procedures, followed by the Toronto physicians and, lastly, those elsewhere in Canada. The distinctions are even more striking for the "perception of seriousness" of certain conditions and, most of all, for "acceptability of abortion." The differences between consecutive groups ranged from 10% to 15%, with Quebec's Anglophone physicians most accepting of abortion, those from

other parts of Canada least accepting, and those from Toronto halfway between.

Quebec's Anglophone physicians thus exhibited a set of attitudes different from that of Toronto physicians who had a comparable practice environment, and from that of other English-speaking Canadian physicians.

A close reading of Table 8.3 will show that Quebec's Anglophone physicians more often resembled their Francophone Quebec colleagues than their counterparts in English Canada. They differed, however, even from Francophone Quebeckers. They saw sterility and sex chromosome anomalies as more serious, while Francophones viewed intellectual impairment and trisomy 21 as more serious. They were much more consistently in favour of selective abortion, even for sex chromosome anomalies. They were less directive and tended to look more favourably on the use of PND procedures (two ultrasound scans per pregnancy, expanded age eligibility for amniocentesis).

In summary, it seems clear that Anglophone and Francophone Quebec physicians are part of the same Quebec culture, distinct from that of other provinces and even from that of Toronto. Quebec's Anglophone physicians also appear to be part of a more technology-oriented, more liberal, and less directive culture. Could it be, as one of our consultants suggested, that they are squarely facing southward (even more so than Francophones), whereas physicians in English Canada are facing eastward or westward?

Conclusion

The 1933 Chicago World's Fair celebrated a century of scientific and technological progress. Its theme was "Science discovers, Industry applies, Man follows." Today, science continues to discover, industry benefits more than ever before from scientific progress, but "man" no longer follows. People are not prepared to follow the dictates of science as blindly as they did in the first third of the century. After Hiroshima, Hitler, thalidomide, and artificial hearts, technology has entered, probably forever, the era of suspicion (Salomon 1991).

This is particularly true in medicine. After World War II, every new technology was perceived as desirable, and public policy sought to encourage its development and dissemination. The focus began to shift in the mid-1960s. Not only were technologies to be developed, but they also had to be made accessible to anyone who needed them, rich or poor, city dwellers or rural folk. In the 1970s, an obsession with costs began to emerge. The question was no longer only whether technology could produce results, but whether its widespread use was truly cost-effective. From the mid-1980s onward, although other issues were far from resolved, a new question arose, namely, whether advances in medical technology

were in keeping with the values and views of the majority of the population. Technological development was henceforth a matter of values as much as effectiveness, cost, or equity.

It took nearly a century for science to free itself from religion and its attendant values and to develop in an essentially scientific way. Fifteen years of spectacular technological progress have now brought medicine back to the type of debate about values that characterized its development before the Flexnerian revolution. As a society, are we prepared to abort any fetus that, in the eyes of medicine, has some form of genetic anomaly? Are we prepared to accept any treatment, whatever the price? Are we prepared to submit to any test solely because it is available and could ease our fears about the future? Are we willing to let our resources go increasingly to medicine and its technology to the detriment of other types of collective investment?

These are the kinds of issues we wanted to deal with in this survey on physicians' attitudes toward PND and its consequences. PND is at the very heart of today's social debate because of its cost, the problem of availability caused by its rapid development, its effectiveness, the scientific value of the various procedures, and also because it touches on moral issues fundamental to our way of being, feeling, and behaving — indeed, fundamental to life itself.

The PND Debate

The birth of malformed or "abnormal" children has often been perceived as a great tragedy for the families concerned. The way Western societies react to this tragedy has evolved considerably (Retsinas 1991). Child abandonment or even infanticide as practised in centuries past are no longer accepted, and systematic institutionalization, with its often disastrous consequences, is no longer common practice. Community resources to assist the parents of disabled children have increased, although probably not enough. But the greatest change is linked to the development of PND.

Before the advent of PND, medicine was ill equipped to help couples known to be at risk. Genetic counselling was based on nothing more than estimations of probabilities. Couples at risk had nothing other than these estimates on which to base their decision whether to have children or not. The advent of ultrasound scanning, amniocentesis, CVS, and blood tests in the 1970s and 1980s changed the situation dramatically. It became possible to say, with some degree of accuracy and certainty, whether a fetus was normal, and thus to decide whether the pregnancy should proceed. People at risk are no longer deprived of children; PND can tell them about the condition of the baby even before it is born and thus relieve many fears. These breakthroughs have opened up a new era of reproductive freedom.

Some of the most vehement criticisms of PND concern the "overmedicalization" of birth and the obstetrical paternalism denounced by feminist movements in the 1970s (Ehrenreich and English 1979; Quéniart 1988; de Koninck 1988). Changes in obstetrical attitudes in the 1980s, the massive number of women entering medicine, and technological development have deflected the thrust of those criticisms in recent years toward a denunciation of the counterproductive, unforeseen effects of advances in PND (Lippman 1991).

These procedures are evolving constantly, becoming safer and less invasive, being performed earlier, and, if more widely marketed in Canada, will become increasingly routine during pregnancy. "Routine" carries an implicit recommendation, one that even the genetic counsellors with the greatest respect for women's freedom of choice cannot get around (Clarke 1991), namely, to abort, if need be. This "need" itself tends to expand (Nelkin and Tancredi 1989) as the number of anomalies that can be diagnosed *in utero* grows and possible indications for PND increase. The technical ability to exercise an ever more intensified qualitative selection of fetuses through the abortion of those that do not possess the desired characteristics opens the door to the systematic elimination of "deviants." Some people fear a new decline in their freedom to choose. They already envisage the possibility that PND, while optional today, might one day become compulsory.

At the same time, there have been changes and controversy in law and judicial practice (Bourgeault 1990). In the past, law and religion were allies in "protecting nature," which was presumed to be the reflection of a divine order. Law then moved away from religion, seeking rather "to protect individual rights" — the right to choose, the right to the best medicine technically possible, etc. Today, law is looking for an identity; individual rights, though important, are not a panacea. A collective morality, still ill defined, is trying to find its way.

In summary, some believe we are on a slippery slope that could lead to disaster. They consider that a radical upheaval in social values lies at the bottom of this slippery slope (increasingly greater acceptability of procedures to assess the condition of the fetus, acceptability of abortion). They fear that, in the medium to long term, a new moral order may become established in the name of progress, characterized by a great intolerance of deficiency and disability, the trivialization of abortion, and the introduction of "productivity" and "efficiency" standards for pregnancy and childbirth.

What This Study Tells Us

The attitude of physicians is important in this context of change, since they are primarily responsible for using and disseminating these procedures, and they are putting into effect the social choices entailed by the procedures. As explained in Chapter 1, this survey was designed to answer three questions: (1) What are the attitudes and behaviours of physicians with regard to the use of PND procedures? (2) How does the medical profession perceive various anomalies that can now be diagnosed prenatally? (3) What is its position on the choices to which the developing technology is leading, given the increasingly large number of "anomalies" that can be diagnosed in utero? In particular, these choices involve the acceptability of abortion, the physician's role in the decision, and the validity of greater government regulation in this area.

Use of Procedures

There is a weak consensus among Canadian physicians (around 60%) in favour of keeping the eligibility threshold for amniocentesis at 35 years of age. Most of those who oppose this norm would raise the threshold. There is no consensus on the eligibility threshold for CVS, and the tendency would be to make access to it more difficult. Of all our respondents, GPs had the greatest reservations in this regard.

As was the case for amniocentesis, there was a weak consensus on the need to perform only one ultrasound scan per pregnancy, with general practitioners at the lower utilization end of the spectrum and specialists at the higher end. Here again, there was a remarkable range of opinion. In Manitoba and Alberta, nearly half the physicians (40%) did not consider it essential to order an ultrasound scan during pregnancy; in Quebec (where the medical group using PND is more specialized), only 4% of physicians shared this opinion. Quebec physicians tended to order two ultrasound scans per pregnancy. Moreover, Quebec was the only province where the great majority of physicians (89% versus 60% for the rest of Canada) considered it acceptable to use ultrasound to screen for anomalies.

Most physicians opposed expanding access to amniocentesis under government health plans for any reason whatsoever (anxiety on the part of the expectant mother, freedom of choice, selecting the sex of the fetus). Contrary to Canadian geneticists' practice guidelines, many physicians (51%) said they do not feel justified in offering amniocentesis to a woman who refuses to consider abortion if an anomaly were diagnosed. On the other hand, they would be prepared to expand access if women paid for the test themselves.

As regards new technological developments, physicians would be prepared to introduce predisposition testing for various common diseases, provided it were used in early childhood or adulthood (not *in utero*). They accepted artificial insemination as a means of preventing the transmission of genetic disorders, and two out of five would accept surrogate motherhood. They were opposed to various procedures that would make it possible to select the sex of a fetus.

Multivariate analysis showed that physicians' attitudes toward various procedures were less conditioned by cultural factors (religion, religious practice, ethnic origin, number of children) than social and professional

characteristics. Apart from the influence of the province where they practised, the more direct contact they had with PND and (unexpectedly) the older they were, the more they tended to favour technological development and the utilization of PND techniques.

Perception of Anomalies

The perception of the seriousness of the various abnormalities listed in the questionnaire varied greatly. Generally speaking, anomalies resulting in a low degree of autonomy (paraplegia, trisomy 21, intellectual deficiency) were perceived as more serious than those suggesting future behavioural problems (e.g., aggressiveness) or fertility problems. But there were broad disparities among provinces. For instance, the majority of physicians in Quebec (70%) said they could not see themselves living with a child with trisomy 21, compared to a minority (40%) in the other provinces (and less than 20% in Saskatchewan). In addition, many more Quebec respondents (more than 84% as opposed to 61% for Canada as a whole) considered intellectual deficiency serious.

Multivariate analyses showed that disparities in how seriously anomalies are perceived are determined more by individual factors than membership in a given group (i.e., sociocultural and professional characteristics). A small part of the variance (10%) was nevertheless attributable to practice area (urban or rural), religion, gender, number of children, province, specialty, and ethnic origin.

Social Choices

The Canadian medical profession unanimously and categorically rejected the use of PND for the purpose of selecting the sex of the fetus. Physicians rejected the idea of utilizing medical techniques for non-medical purposes.

Fifteen percent of Canadian physicians were opposed to abortion following diagnosis of an anomaly, no matter what the anomaly might be. This figure was surprising, since in our previous surveys (France and Quebec) the percentage of physicians unconditionally opposed to abortion was never more than 5%. The remaining physicians, who were not unconditionally opposed to abortion in the case of an anomaly (85%), were distributed along a normal curve ranging from opposed in some circumstances to extremely sympathetic.

Given the historical prominence of trisomy 21 as justification for the development of amniocentesis, we expected that a majority of physicians, as in our previous surveys, would accept abortion for this anomaly. Fifty percent of Canadian physicians were receptive to abortion for trisomy 21 without structural malformations, with extremely pronounced regional disparities (ranging from 25% in Saskatchewan to 70% in Quebec). In this respect, Quebec's Anglophone physicians (the group that was by far the most open to selective abortion for all kinds of anomalies) constituted a distinct group within Canada.

The percentage of physicians favourable to abortion for the other listed anomalies was, as might be expected, even lower than for trisomy 21. Generally speaking, religion, religious practice, specialty, ethnic origin, and province of practice were the best predictors of abortion acceptability. These variables, plus the perceived seriousness of various problems, accounted for more than 30% of the variance (as much as 60% in some provinces).

Similarly, four out of five Canadian physicians objected to the eugenic statements contained in the questionnaire. The data suggest that the more general the practices of physicians, the more they are opposed to any form of control designed to eliminate anomalies. Conversely, the more technically oriented the practices, the more the physicians perceive it as unacceptable to give birth to a malformed child when the anomaly can be diagnosed, and the more they measure the success of PND in terms of the costs avoided by abortion. Only a minority — 28% of Canadian physicians — were of that opinion, however.

No consensus exists in Canada on whether it should be left entirely to parents to decide on abortion (50% in favour, 36% against). There was a weak consensus, however, that physicians should sometimes intervene in the parents' decision, in particular to oppose abortion where anomalies are considered minor. A number of doctors (between 16% and 63%, depending on the item) consider it part of their role to offer direction with regard to the decision to abort. However, while they sometimes find it difficult to disclose all the information they have to the parents, all physicians feel an obligation to do so, with the exception of fetal sex (37% opposed with regard to CVS).

Lastly, a majority of physicians surveyed (62%) accept existing regulations on the eligibility age for amniocentesis. They are prepared to widen access to PND, but only if patients make a direct financial contribution. They also agree that government should invest additional resources if genetic predisposition tests become available. On the other hand, they gave greater priority to funding preventive social programs (prevention of low birthweight, anti-smoking campaigns, etc.) than to development of genetic screening technology.

Are Concerns About PND Justified?

Two characteristics emerge from this "family portrait" of the Canadian medical profession. The first one is that physicians are just as divided in their opinions as all Canadians undoubtedly are. What is most striking about our findings is not so much the consensus — there were several, such as the obligation to disclose all information — as the diversity of opinion on such important issues as technological development, the seriousness of various anomalies, the conditions under which abortion is acceptable, and the role of the physician. This is somewhat reassuring, since technological development does not seem to have made social

relations more homogeneous, at least for the time being. multivariate analysis shows, when deciding on a difficult question physicians rely not so much on the dictates of medical science (as reflected in their medical school, the distance to the nearest genetics centre, or their professional environment) as on the deep-seated values they hold from their membership in primary groupings such as province, specialty, ethnic group, and religious practice. In this respect, they are no different from the average citizen: they do not know where PND is leading. It is their culture, in the sociological sense of the word, that colours their attitudes.

The second characteristic of the medical profession is its cautious and conservative approach to PND. Faced with the concerns raised by PND. physicians currently either favour the status quo (e.g., opposition to using PND to select the sex of the fetus or to relieve a patient's anxiety) or are ambivalent (e.g., number of ultrasound scans per pregnancy, eligibility age for amniocentesis, abortion acceptability). They are neither enthusiastic about eugenics nor overly fascinated by the possibilities of today's technology. They also tell us that their preference for combatting birth defects would be the funding of preventive social programs rather than technology.

At first sight, these characteristics appear reassuring. The "system" seems to be subject to just and efficient regulation. With such debate and heterogeneity among Canadian physicians, one might expect the future evolution of PND to continue to reflect the diversity of Canadian society.

However, a closer look reveals some puzzling results.

The attitudinal disparities among provinces, particularly between Saskatchewan at the most conservative end of the spectrum and Quebec at the most liberal, are very pronounced. To judge from our data (see Figure 8.3 in particular), the medical fate of Canadians varies drastically according to where they live in Canada, a country that has made equitable access to equal quality services a basic tenet of its public health system. As this study suggests, and as the study by Hamerton et al. (1993) demonstrates for amniocentesis, access to PND is very unequal throughout the country. There are also broad variations between provinces with respect to how serious certain conditions are considered to be. how directive physicians are, and how readily they accept selective abortion.

The divergence of opinion with regard to perception of seriousness and the variance in the perceived reliability of ultrasound scanning for diagnosing fetal anomalies are also intriguing. Might there be differences in knowledge between physicians that training programs could correct? How can we account for the rather surprising differences in perception of the seriousness of anomalies, such as intellectual impairment and trisomy 21? How can physicians say they have no faith in ultrasound scanning for diagnosing anencephaly? Why do some consider it unjustified to use ultrasound to screen for malformations? Why do some reject the use of ultrasound during pregnancy when the majority consider it a routine procedure? It could be that their opinions stem from specific personal experiences. It could be that these points of view are based on the valid

interpretation of various evaluation studies. But it could also be that the competence of some physicians is in question. Unfortunately, this study was not designed to measure this aspect.

The discrepancy between age groups was also striking when compared to other studies (e.g., Julian et al. 1989b). On the one hand, younger physicians did not differ from their elders in their acceptance of abortion. On the other hand, the younger a physician was, the more wary he or she was about using the procedures and technological development. Is this an attitude that carries implications for the future, or is it attributable to a lack of perspective, passing fads, or a blasé attitude toward technology among young physicians who have lived all their lives in a technically supercharged environment?

Most physicians opposed expanding PND procedures for purposes that are not strictly medical (anxious mother, choosing sex of fetus, etc.). They also opposed expanded access to amniocentesis within the public health care system. On the other hand, when asked if they would be prepared to order amniocentesis for no other reason than that the woman was willing to pay for it (assuming that regulations would permit it), most said yes. Does this mean that, given different financial incentives, physicians would reappraise their present cautious and prudent stand?

The Future

No doubt the Canadian medical profession's cautious and contradictory stance can be explained by the present state of knowledge, technology, and regulations regarding PND. Should this situation change, the attitude of the medical profession will likely change as well.

First, most of the procedures used today are invasive (amniocentesis, CVS) and done rather late in pregnancy, and thus are risky. Ultrasound scanning is not yet fully reliable in detecting malformations, particularly in early pregnancy, and blood testing is still experimental. If this situation were to change, physicians would no doubt be more inclined to use the tests.

Second, as we have seen, when taken to its conclusion PND can result in malformed fetuses being aborted. This causes much uneasiness and a host of moral problems. If treatment became available for certain anomalies, physicians would no doubt be more enthusiastic about PND. Current research on the human genome, genetic engineering, and advances in fetal surgery could make treatment a reality sometime in the not too distant future. Should it happen, we may assume that there will be less conflict between physicians' values and their perception of PND. There would be fewer moral constraints on its spread, although other problems (stigmatization, job discrimination, etc.) would continue to give cause for moral concern.

Third, changing the way PND is funded could also lead to a change in attitude. The Canadian public health system currently prohibits extra

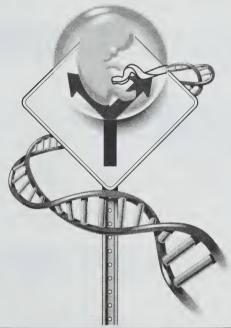
What does the future hold? Considering the uncertainty about how the state of knowledge, technology, and regulation will evolve, it is difficult to make reliable predictions. One thing is sure: PND is here to stay. Equally sure is the fact that it entails some upheaval in social values, as can be seen from the newly acquired legitimacy of *in utero* testing and selective abortion. In the meantime, guided by their personal values, Canadian physicians overall have maintained a restrained and cautious attitude that, in most parts of the country, has produced a wide variety of individual choices.

It is obviously impossible to say whether technology will someday make social relations totally homogeneous or whether, after spreading slowly, PND will drag us down a slippery slope that will change for the worse humankind's way of looking at deficiencies, pregnancy, and abortion. The door remains open to intolerance, stigmatization, arbitrariness, and eugenics. But living in society implies living with a large number of "slippery slopes" and difficult choices; it is how we adjust to them that defines our society. Despite its major limitations (a questionnaire format that provided only a superficial look; study of physicians only, not women), what this study seems to tell us is that there is no reason to worry at present, provided, of course, that we remain vigilant and that PND techniques are increasingly subject to rigorous evaluation with regard to both their effectiveness and their social impact.

Appendix 1. "Prenatal Diagnosis at the Crossroads": A Survey of Canadian Physicians

"Prenatal Diagnosis at the crossroads"

A survey of Canadian physicians.



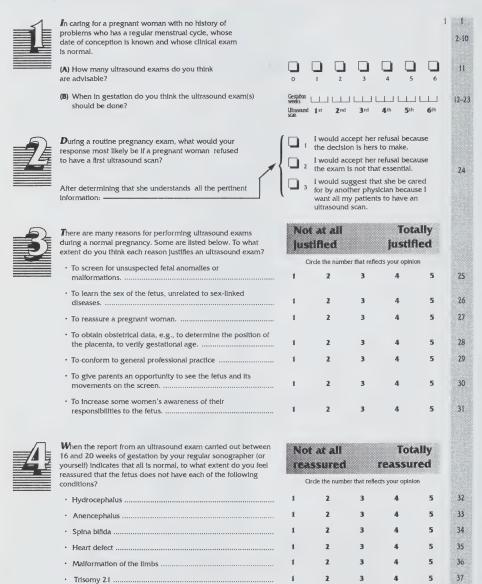
This questionnaire is exclusively intended for the following categories of physicians. Please indicate whether you are:

- an obstetrician-gynecologist with an active obstetrical practice
- a general practitioner/family practitioner performing 5 or more deliveries per year
- a radiologist performing 100 or more obstetrical ultrasounds per year
- a pediatrician

If you belong to one of the above categories, please answer the questionnaire.

If not, please return it unanswered in the enclosed envelope.

Your opinions about the use and practice of prenatal diagnosis.





Irrespective of present policies, at what age do you think antenatal screening for fetal chromosomal anomalies should be offered to women who have no family history of problems? [Ctrcle one answer for each technique]

	nswer for each tech									
(A) Amnlocer	ntesis	At all ages	<30 3	0 31	32	33	34 35	36 37 3	8 39 ≥4	40 Neve
		1	2 3	4	5	6	7 8	9 10	11 12 1	3 14
(B) Chorionic	villus sampling	At all ages	<30 3	0 31	32	33	34 35	36 37 3	8 39 ≥4	40 Neve
		1	2 3	4	5	6	7 8	9 10	11 12 1	3 14
	ld woman is pregnan				1	٠,	I would	l advise he	r to be test	ted.
	to conceive. She is s, and does not want					 2	I would	i advise he	r not to be	tested
	any way. At the sam child with trisomy 2		oes not	want to	1	<u> </u>	ultrasou	l advise her und exam to or amniocen	o determin	thorough e the
	ing the risks of testing					п.	Other		itesis.	
	with trisomy 21 with likely to behave?					4	Other	(spe	edfy)	
					(٦,	Lwoule	l advise he	r to be test	ned.
If this woman to behave?	were 38 years old, I	how would ye	ou be mo	ost likely	y [] 2		l advise he		
to believer =					- [] 3	ultrasou	advise her and exam to	o determin	horough e the
						٦.		r amniocen	itesis.	
					(4	Other .	(spe	dfy)	
	request prenatal di	agnosis for r					t at al		To	tally
			- 6 48							
tests. Please o	ose usually consider consider each case de t you find the reason	scribed belov	v, and in	dicate			eptab		accept	able
tests. Please of to what extent 1.A couple is child. The v sea. She is	consider each case de t you find the reason s very worried about woman says she has 33 years old and has	scribed below for the reque giving birth to insomnia and s no history of	v, and in st accept o an abn frequen f genetic	dicate table. ormal t nau- disor-		acı	ceptab		accept	
tests. Please of to what extent 1.A couple is child. The visea. She is ders, but ferecently ga	consider each case de t you find the reason s very worried about woman says she has	scribed below for the reque giving birth to insomnia and is no history of cause a 30-yea lith trisomy 21	v, and In st accept o an abn frequen f genetic ar-old frie . They i	dicate table. ormal t naudisordend request		acı	ceptab	le .	accept	
tests. Please of to what extent 1.A couple is child. The visea. She is ders, but for recently ga amniocent 2.A couple reold. They is the state of	consider each case de t you find the reason s very worried about woman says she has 33 years old and has eels very anxious becave birth to a child wi	giving birth to insomnia and is no history or ause a 30-yeath trisomy 21 casts. The worn I not terminate	o an abn frequen f genetic ar-old frie They nan is 39	dicate table. ormal t naudisordend request years		acı	Ceptab	le .	accept	nion
tests. Please of to what extent 1. A couple is child. The vesses. She is ders, but for recently geamnlocent 2. A couple model. They if the fetus 3. A 33 year-knowledge her age the rather small	consider each case de tyou find the reason severy worried about woman says she has in 33 years old and has eels very anxious becave birth to a child witesls. —equests amniocente state that they would has an anomaly. —old woman requests be tisked of having a child. II. However, since pre	escribed below for the reque giving birth to insomnia and is no history of ause a 30-yeath trisomy 21 to the trisomy 21 to terminate of the test, and fill with a trisoental tests especially tests especially the test of the test, and fill with a trisoental tests especial tests especially tests e	v, and In st accept o an abn I frequent f genetic They I from I for the properties. They I for the properties of the pro	dicate table. ormal t naudisordend request years egnancy that at the believes	, , ,	acı	Circle the nur	The company of the co	accept	alon 5
tests. Please of to what extent 1. A couple is child. The vesa. She is ders, but fe recently ge ammlocent 2. A couple reold. The vest if the fetus 3. A 33 year-knowledge her age the rather small it is for her the risks she	consider each case de tyou find the reason is very worried about woman says she has! 33 years old and has eels very anxious becave birth to a child wites! equests amniocente state that they would has an anomaly old woman requests eable about the risks of risking a climate of the risks of having a climate of the risks of the risks of the risks of having a climate of the risks of the ris	escribed below for the reque giving birth to insomnia and is no history of ause a 30-yealth trisomy 2.1 the trisomy 2.1 to the trisomy 2.1 to the trisomy 3.1 to the trisomy 4.1 to the	v, and In st accept on an abn if requent genetic ar-old file. They in an is 39 to the product of the state of of th	dicate table. ormal t naudisordend request years egnancy is a that at the believes choose		acı	Circle the nur	The company of the co	accept	nion 5
tests. Please of to what extent 1. A couple is child. The vest is ders, but for recentity ge amniocent 2. A couple in old. The vest if the fetus 3. A 33 year-knowledge her age the rather small it is for her the risks sh 4. A couple halready have tally planning attent in the proper in the proper is seen to the couple halready have tally planning attent in the proper is seen of the couple halready have tally planning seen of the couple halready have tally planning seen of the couple halready have tally planning seen of the couple halready have seen of the couple halready have tally planning seen of the couple halready have tally planning seen of the couple halready have the couple halready have tally planning seen of the couple halready have the couple halready have the couple halready have the couple hall the couple	consider each case de tyou find the reason is very worried about woman says she has is 33 years old and has eels very anxious becave birth to a child witesis. "equests amniocente state that they would has an anomaly	scribed below for the reque giving birth to insomnia and in on history of ause a 30-year. Ith trisomy 21 in the trisomy	v, and In st accept on an abn frequent of genetic arrold frid. They in an is 39 to the process. She disperse the process of th	dicate dicate table. ormal fr naudisor-end eequest years egnancy. Is it that at re believe choose years an initiate table.	, , , , , , , , , , , , , , , , , , ,	acı	ceptab Circle the nur 2 2	The company of the co	accept	nion 5



 \boldsymbol{W} hen fetal karyotypes are examined during the course of prenatal diagnosis for women 35 years and over, chromosomal abnormalities such as XYY Syndrome*, Klinefelter syndrome**, and Triple X syndrome*** may be detected.

Extremely Not at all severe severe Circle the number that reflects your opinion

(A) Given the current state of knowledge, what is your assessment of each syndrome?

 XYY syndrome

5 3 5 2 3

AR.

50

51

52

53

(B) If you were not legally required to do so, would you consider it appropriate to tell the parents that the fetus carries one of the following syndromes? XYY syndrome

YES NO

Triple X syndrome

*XYY SYNDROME: The XYY syndrome occurs in about 1.5 per 1000 newborn male infants. Although more prevalent among inmates of high security Institutions, this syndrome is less strongly associated with aggresive behavior than previously thought, and many affected males remain undetected clinically. Mild mental retardation and behavioral problems can occur, and tall stature is usual. (British Medical Journal, 1989, vol. 289)

"KLINEFELTER SYNDROME: The XXY karyotype of Klinefelter syndrome occurs with an incidence of 2 per 1000 liveborn males. The primary feature of the syndrome is hypogonadism, and affected males are usually tall. Pubertal development often progresses normally, but testosterone replacement treatment is sometimes required. Testicular size decreases after puberty, and affected males are Infertile. Gynaecomastia may occur, and the risk of cancer of the breast is increased. Intelligence is generally within the normal range, but educational difficulties and behavioural problems are fairly common. (British Medical Journal, 1989, vol. 289)

"TRIPLE X SYNDROME: The triple X syndrome occurs with an incidence of 0.65 per 1000 liveborn female infants. Apart from being taller than average, affected girls are physically normal. Educational problems are encountered more often in this group than in the other types of sex chromosomal abnormalities. Mean intelligence quotient is lower than in controls, about half of affected girls having delayed speech development and three quarters requiring some remedial teaching. Gonadal function is usually normal but premature ovarian failure may occur. (British Medical Journal, 1989, vol. 289)



1	2	3	4	5	54
1	2	3	4	5	55
1	2	3	4	5	56
1	2	3	4	5	57
1	2	, 3	4	5	58
1	2	3	4	5	59
1	2	3	4	5	60
i	2	3	4	5	61
1	2	3	4	5	62
1	2	3	4	5	63
		1 2 1 2 1 2 1 2 1 2 1 2 1 2 1 2 1 2	Circle the number that reflect 1	Circle the number that reflects your opinion in the circle and the	Circle the number that reflects your opinion 1

Not at All

77 78

79

80

81

82 83 84

85

86



For each of the following conditions diagnosed in a fetus, please Indicate the extent to which you believe pregnancy termination is acceptable.

	**************************************	ircle the num
A very severe heart defect that will require several surgical operations after birth.	1	2
Lobster claw defect of the hand	1	2
Spina bifida without evidence of hydrocephalus	i	2
Trisomy 21 without evidence of structural malformations	1	2
XYY syndrome	1	2
Klinefelter syndrome (XXY).	1	. 2
Triple X syndrome.	1	2
Turner syndrome (XO)	1	2
Cystic fibrosis.	1	2
Duchenne muscular dystrophy	1	2
Huntington disease.	1	2
Phenylketonuria	1	2
A fetus of the non desired sex.	1	2

	No	t at All		Tot	ally	
	acc	ceptabl	e a	ccepta		
		ircle the numb				
	1	2	3	4	5	64
	1	2	3 "	4	. 5	65
	1	2	3	4	5	66
	1	2	3	4	5	67
	1	2	3	4	5	68
	1	. 2	3	4	5	69
,	1	2	3	4	5	70
	1	2	3	4	5	71
	1	2	3	4	5	72
	1	2	3	4	5	73
	1	2	3	4	5	74
	1	2	3	4	5	75
	- 1	2	2		E	76



The field of molecular genetics is expanding rapidly. With these new techniques, an increasing number of predispositions to a wide range of conditions will be diagnosable in utero, at birth or shortly after, or at any stage in life. These tests for predispositions will identify individuals with a genetic susceptibility to a condition, but they cannot predict with certainty whether he or she will develop that condition and at what stage in life. Research in this area is in progress..

(A)	If tests for predispositions to the conditions below became
	available, at what stage in life do you think they should be
	done? (Check only one answer for each condition)
	· Diabetes
	: Alcoholism

(B)	Which reason would	Justify	testing for genetic	

Coronary heart disease

· Schizophrenia

· Coronary heart disease

predispositions to the conditions below? (Check only one answer for each condition)

Alzhelmer's disease

· Dlabetes
· Alcoholism
· Schlzophrenia
Alzheimer's disease

in utero	or early infancy	hood	Never
1	2	3	4
To prevent births	To provide early treatment 2	To provide preventive coun- selling	None 4

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The allocation of financial resources for health care poses difficult problems for the medical profession. If you were planning a new budget for health care, how would you allocate the money?

[Please rank in order of importance from 1 (most important) to 7 (least important). Use each	number only ones.
To support multidisciplinary teams that will provide care to socially disadvantaged	oregnant women
• To increase the budget and improve the services of cytogenetic laboratories	
· To Improve training in obstetrical ultrasonography and increase the number of spe	ialists doing scans
 To Implement population-wide prenatal screening blood tests (e.g., maternal serur tests) to identify for prenatal diagnosis women at genetic risk (trisomy 21, spina bil their age. 	AFP, hCG, oestriol da), irrespective of
To develop integrated nutritional assistance and counselling programs for women reduce the number of bables with low birth weight.	t risk in order to
To develop wide-scale information programs about the harmful effects of alcohol a during pregnancy.	nd cigarettes
To develop services for the treatment of infertility	
$\emph{\textbf{I}}_{ ext{n}}$ your opinion, how important is the assessment of exposure	Not important Extremely at all important 1 2 3 4 5



to mutagenic and teratogenic hazards (e.g. radiation and chemicals) In a prenatal questionnaire?

00000

Your OPINION ON SOCIAL QUESTIONS IN REGARDS TO PRENATAL DIAGNOSIS.



Prenatal diagnosis* raises many ethical and medical questions in every country in the world. Listed below are statements that reflect a variety of medical, scientific, religious, and political points of view. What is your opinion?

* We include as prenatal diagnosis techniques such as amniocentesis, obstetrical ultrasonography, chorionic vilius sampling and several prenatal screening blood tests.

	Tot	ally attre			tally				ally agree			tally gree	
I sometimes feel legally bound to reveal information to parents that I would prefer to withold.		2	3	4	gree 5	95	In carrying out their work, physicians must not consider the costs of medical services.	í	2	3	4	5	105
A physician must be able to resist some abortion requests when he or she considers an anomaly to be minor	1	2	3	4	5	96	The abortion of a fetus with a minor malformation is acceptable Women having obstetrical ultrasound scans	1	2	3	4	5	106
 There is a danger that results from tests for genetic predispositions will be used for discriminatory purposes. 	1	2	3	4	5	97	should be asked for their written consent before being examined.	1	2	3	4	5	107
With respect to abortion, parents have an absolute right to freedom of choice	1	2	3	4	5	98	14. Organized groups for the handicapped should be consulted during the development of policies with respect to prenatal diagnosis.	1	2	3	4	5	108
A woman who does not meet general poll- cy guidelines for an amniocentesis should have access to the test if she is willing to pay the costs of this service herself	1	2	3	4	5	99	15. It is useless to test for genetic predispo- sitions to conditions that cannot now be treated.	1	2	3	4	5	109
Physicians, not parents, should decide which fetal anomalies warrant pregnancy termination.	1	2	3	4	5	100	16. It is acceptable to suggest the option of "surrogate motherhood" to couples when the woman has an autosomal dominant disorder.	1	2.	3	4	5	110
With increasing refinement in the techniques for prenatal diagnosis, conditions which we would otherwise consider "normal" and accept as just a part of life are now seen as pathological.	1	2	3	Δ	5	101	 Abortion on request is less acceptable than abortion of a fetus with an anomaly. 	1	2	3	4	5	111
The success of prenatal diagnosis is best measured by reductions in the costs of ser-	,	_	,	•			18. In general, women rely on prenatal diagnosis too much.	1	2	3	4	5	112
vices for the care of children with genetic anomalies.	1	2	3	4	5	102	19. Use of prenatal diagnosis makes us more and more intolerant of the smallest anomaly in a fetus or child	1	2	3	4	5	113
The use of techniques for preconceptional determination of fetal sex (chromosomal selection) is acceptable.	1	2	3	4	5	103	20. A physician should not tell parents about a fetal anomaly if he/she considers it a minor anomaly	1	2	3	4	5	114
10.One must condemn prenatal diagnosis done with the deliberate intention of having an abortion if the results reveal							21. Abortion of a fetus with an anomaly is more justifiable in the first trimester than						
the existence of an anomaly	1	2	3	4	5	104	in the second trimester of pregnancy	1	2	3	4	5	115

22. It is acceptable to suggest the option of artificial inserimation to couples when the man has an autosonal dominant disorder. 1 2 3 4 5 116 23. In some cases, a physician must influence the parents' decision to confinue or to abort a pregnancy. 1 2 3 4 5 117 24. A physician must discuss the question of abort a pregnancy. 25. The sale of simple self-prescribed tests to identify fetal sex should be allowed. 25. The sale of simple self-prescribed tests to identify fetal sex should be allowed. 26. The sale of simple self-prescribed tests to identify fetal sex should be allowed. 27. A minimum only 3% of children are born with third feders while a must large proportion born in good health develop setious bandcians caused by social or economic conditions. 27. A minimum on the first rage, or martial of socioeconomic status. 28. Chirty pitch intentionally to a child with a genetic decist at mine when both premated diagnosis and abortion are available to a genetic decision to confinue their pregnancy so that the fetus's exhallent pregnancy so that the fetus's healthy or acceptable fetals to confinue with an amenaphalic fetals to confinue their pregnancy so that the fetus's healthy or acceptable fetals to confinue their pregnancy so that the fetus's healthy or acceptable fetals to confinue their pregnancy so that the fetus's healthy or acceptable fetal to confinue their pregnancy so that the fetus's healthy or acceptable fetals to confinue their pregnancy so that the fetus's healthy or acceptable fetals to confinue their pregnancy so that the fetus's healthy or acceptable fetals to confinue their pregnancy so that the fetus's healthy or acceptable fetals to confinue their pregnancy so that the fetus's healthy or acceptable fetals to severe the fetals to the score fetals the fetus's healthy or acceptable fetals to the score fetals the fetus's pregnancy so the fetals the fetus's pregnancy so that the fetus's pregnancy so the fetals the fetus's pregnancy so the fetals the fetus's pregnancy so the fetals the fet	Question 15	continued		ally agre			tally gree		Totally Totally disagree agree
23. In some cases, a physician must influence the parents' decision to continue or to abort a pregnancy	of artificial when the	Insemination to couples man has an autosomal domi-							28. Giving birth intentionally to a child with a genetic defect at a time when both prenatal diagnosis and abortion are
24. A physician must discuss the question of abortion with alcoholic women	23. In some ca	ases, a physician must influence is' decision to continue or to	1	2	3	4	5	116	29. It is acceptable to encourage women with an anencephalic fetus to continue
25. The sale of stmple self-prescribed tests to ledentify fetal sex should be allowed			1	2	3	4	5	117	healthy organs may be used for
26. Prenatal diagnosis annot be considered a priority when only 3% of children are born with birth defects while a much larger proprotion born in good health develop serious handlcaps caused by sodal or economic conditions. 1 2 3 4 5 31. Farer of lawsuits makes us use prenatal diagnosis more than is necessary. 1 2 3 4 5 32. Farer of lawsuits makes us use prenatal diagnosis more than is necessary. 33. Farers having chorolic villus sampling should not be given information on fetal sex unless it is medically relevant. 33. Farers having chorolic villus sampling should not be given information on fetal sex unless it is medically relevant. 34. If it were possible to identify all cystic fibrosis carriers, a systematic detection of the soft provided	abortion w	vith alcoholic women	1	2	3	4	5	118	
a priority when only 3% of billiden are born with birth defects while a much larger proportion born in good health develop serious handicaps caused by social or connomic conditions			1	2	3	4	5	119	control the spread of genes causing
larger proportion born in good health develop serious handicaps caused by social or economic conditions	a priority v	when only 3% of children are							32. Fear of lawsults makes us use prenatal
social or economic conditions	larger proj	portion born in good health							33. Parents having chorionic villus sampling
### Country/province/state where the diploma was obtained: What is your professional specialty? Check one answer or more)	27. Amniocen	tesis should be made available	1	2	3	4	5	120	34. If it were possible to identify all cystic fibrosis carriers, a systematic detection of
## Attended medical school Medical school from which you received your medical degree: Medical school from which you received your medical degree: NF			1	2	3	4	5	121	
• general practitioner		was obtained:	spec	lalty	1?		_	4-5	Area In which you practice:
* radiologist		• family practitioner • obstetrician-gynecologist performing deliveries -chie	effy		((1 2 3			• less than 100 km 1 • between 100 and 250 km 2
other (specify) Type of practice: (check one answer or more) solo practice collectrice practice -2 physicians 3 10 -4 physicians or more 4 11 -4 thought 12 -4 thought 13 -4 thought 14 thought 14 thought 14 thought 15 -4 thought 15 thought 16 though 16 thought 16 though 16		radiologist pediatrician ★(neonatalogy) -ex -occ	clusiv	ely .	((] 6] 7] 8		6-7	Language spoken with the majority of your patients: (Check one)
Type of practice: (check one answer or more) solo practice — 2 physicians — 3 10 4 11 4 10 10 10 12 12 12 12 10 12 12 12 12 10 12 12 12 12 12 12 12 12 12 12 12 12 12		• other	/er		[
*solo practice		_	uns we	r or m	ore)				
-6 physicians or more 4 11 did you perform	TO	· collective practice -2 physician	15		Č	2		9	· deliveries did you perform
annual de 12		-6 physician	s or r	nore	Ü	4		11	dld you perform
-general			******						new-poins did you care for

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	$oldsymbol{W}$ ho performs obstetrical ultrasound exams	(white	D	MAKK
	for your clients?		ERSONAL INFORMATION	
	(Check all appropriate answers)	20	S ex:	
	· an obstetrician 1	33	· man	
	• a radiologist 2	34	· woman	64
	other3	35	· woman	
	(specify)			
	During the past year, approximately		Year of birth:	
34	how many of the pregnant women in			65-66
1 5 差	your practice had:			
	· amniocentesis (number)	36-38		
	· chorionic villus sampling (number)	39-41	Number of children:	
	maternal serum	45.5		
	alpha-fetoprotein tests (number)	42-44		67-68
	Which percentage had an obstetrical ultrasound	45 47		
	obstetrical ultrasound%	45-47	Mother tongue:	
50	What is the socio-economic status of most		english	
	of the patients in your practice?		• french	69
		48		37
	upper class	49	· other 3	
	· lower class	50		
	• other 4	51	To which ethnic or cultural group(s) did your parents (mother and father) belong?	
	(specify)		your parents (mother and lather) belong?	
~_			(For example: German, English Chinese. French.	70-73
	What is your main source of information		Greek, Haltian, Inult, Italian, Japanese, Lebanese,	
3 4 E	In following new developments in prenatal diagnosis?		Polish, Ukrainian, etc.)	
		44	What is your religion?	
	· conferences	52		74-75
	· continuing medical education 2	53	(For example: None, Anglican, Baptist, Buddhist,	
	• scientific Journals	55	Greek Orthodox, Hindu, Islam, Jehovah's Witness, Jewish, Lutheran, Mennonite, Pentecostalist,	
	· colleagues	56	Presbiterian, Roman Catholic, Salvation Army, Sikh,	
	• medical journals	57	Ukrainian Catholic, United Church)	
	• other	58	D o you practice your religion?	
	(specify)	30	• yes	
			· sometimes 2	76
-	In your professional activities, do you consider		· no	
7/0	yourself to be: Not at all Totally			
	· directive in your advice		Your comments about this study	
	to patients	59	Did you find:	
	· an early adopter of	60	• the subject interesting 1 2 3 4 5	77
	new technologies		• the questions clear	78
	sional opinions with colleagues	61	the research relevant	79
	likely to discuss your opinions with patients	62	the size of the questionnaire	80
A	In most aspects of your non-professional life,		THANK YOU	
	do you consider yourself		Your cooperation and contribution to this study are	
	• more conservative than liberal		greatly appreclated.	

· equally conservative and liberal

· more liberal than conservative

Marc Renaud and colleagues

Prenatal Diagnosis at the crossroads Tel.: (514) 343-6193

Appendix 2. Categorization of Religions and Ethnic Origin

Categorization of Religions Catholic - Roman Catholic - Ukrainian Catholic Anglican - Anglican - English Catholic United - United Church Church Protestant - Christian Reformed - Lutheran (others) - Baptist - Mennonite - Brethren - Methodist - Christian United Church - Moravian - Evangelical - Seventh Day Adventist - Evangelical - Presbyterian - Quaker - Unitarian - Salvation Army - Religious Society of Friends Judaic - Judaic - Hebraic Oriental - Islamic religions - Buddhist - Hindu - Jain - Sikh No - Agnostic religion - No religion Others - African - Christian Science - Alliance - Latter Day Saints - Missionary Alliance - Jehovah's Witnesses - Other - Long House - Baha'i - Mormon - Christian - North American Indian - Coptic - Nazarene - Generic - New Age - Greek Orthodox - Orthodox - Quiescent Hedonist - Humanist - Ukrainian Orthodox

- Universalist

- Jrez Tminken

Categorization of Ethnic Origin

English

- American
- English
- Australian
- English Canadian
- Scottish
- Irish

French

- Acadian
- Belgian
- French Canadian
- French
- Québécois

European (other than British or French)

- German
- Austrian
- Celtic
- Danish
- Spanish
- European
- Finnish
- Greek
- Icelandic
- East European
- Bulgarian
- Caucasian - Croatian
- Estonian
- Hungarian
- Latvian
- Lithuanian
- Macedonian
- Polish
- Jewish
- Jewish
- Asian
- Asiatic
- Burmese
- Chinese
- Korean - Hindu
- Hilliuu
- East Indian

- ItalianMaltese
- Norwegian
- Portuguese
- Scandinavian
- Swedish
- Swiss
- Welsh
- Rumanian
- Russian - Slovak
- Slovene
 - 01040110
- CzechoslovakianUkrainian
 - Oktailliali
 - Yiddish
 - Yugoslavian
 - Japanese
 - Malaysian
 - PakistaniFilipino
 - O : I
 - Sri Lankan
 - Vietnamese

Appendix 3. Frequency Tables

Chapter 3. Sociocultural and Professional Profile of Physicians

A 3.1. Physician Population Under Study, by Specialty, Number of Respondents, and Response Rate for Canada as a Whole

	Population	Sample	Number of respondents	Response rate (%)	Weighted number of respondents
GPs	8 021	1 715	1 045	60.93	1 961.82
Obstetricians	1 528	1 501	773	51.33	372.75
Paediatricians	2 027	1 746	770	44.10	495.02
Radiologists	991	991	484	48.84	242.37
Total	12 567	5 953	3 072	51.60	3 071.96

A 3.2. Physician Population Under Study, Number of Respondents, and Response Rate by Province

	Population	Sample	Number of respondents	Response rate (%)	Weighted number of respondents
Atlantic	1 244	489	240	49.08	303.5
Quebec	1 943	1 193	738	61.60	474.73
Ontario	4 085	2 461	1 249	50.75	999.15
Manitoba	624	310	151	48.71	152.7
Saskatchewan	588	177	88	49.72	143.1
Alberta	1 445	551	253	45.92	353.7
British Columbia,					
NWT, and Yukon	2 638	772	353	51.56	645.9
Total	12 567	5 953	3 072	51.56	3 071.96

· /								
	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
GPs	222	194	569	98	112	256	510	1 962
Obstetricians	26	98	155	16	10	29	39	373
Paediatricians	30	141	186	32	12	43	51	495
Radiologists	26	42	89	7	10	25	45	242
Total								
population	304	475	999	152	144	353	645	3 072

A 3.4. Male and Female Physicians, by Province (%) (Q30)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
	73.0		76.2 23.8	74.2 25.8		72.2 27.8	76.2 23.8	76.2 23.8
remaie	27.0	21.7	20.0	25.0	,0.0	27.0	23.0	(6)*

^{*} The number of non-respondents appearing in parentheses in Tables A 3.4 to A 3.30 applies to Canada as a whole.

Note: Figures in bold type are statistically significant at $p \le 0.1$.

A 3.5. Age of Physicians, by Province (%) (Q31)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Under 35	23.0	12.0	12.5	19.9	11.3	14.8	10.8	13.7
35-39	23.3	19.3	23.0	23.0	19.1	29.6	22.5	23.0
40-49	38.0	37.6	35.9	35.7	36.8	40.9	45.0	38.9
50-59	11.0	19.3	18.1	11.5	16.9	8.6	14.8	15.4
60 and over	4.6	11.7	10.4	9.9	15.8	6.1	6.8	9.0 (23)
Mean age	41.6	45.7	44.8	43.1	46.8	42.1	43.9	44.1

A 3.6. Number of Children of Physicians,	by Province (%) (Q32)
--	-----------------------

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
0 1 2 3 and more	13.2 10.1 33.6 43.1	14.7 10.7 29.8 44.8	10.5 9.4 31.7 48.4	13.9 5.9 33.6 46.6	6.4 7.4 33.2 52.9	13.9 7.0 35.5 43.7	16.1 4.1 23.4 56.4	12.9 8.0 30.4 48.6 (38)
Mean number of children	2.3	2.3	2.4	2.4	2.6	2.4	2.5	2.4

A 3.7. Mother Tongue of Physicians, by Province (%) (Q33)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
English French Other	94.2 2.9 2.9	16.6 75.6 7.8	83.7 2.8 13.5	83.9 4.7 11.4	69.6 5.4 25	86.0 2.1 11.8	86.6 0.6 12.8	74.5 13.8 11.6 (23)

A 3.8. Religion* of Physicians, by Province (%) (Q35)

	ATL	QUE	ONT	MAN	SASK ALB	BC/NWT	CAN
Roman Catholic	17.7	79.5	21.2	17.6	25.2 18.6	15.9	28.6
Greek Orthodox	0.3	1.2	1.1	0.4	3.4 2.5	0.1	1.0
Anglican	16.3	-	12.1	8.4	6.7 7.7	14.1	10.1
United Church	20.9	-	15.6	19.7	10.7 15.3	14.3	13.3
Protestant	14.4	6.3	14.7	24.3	33.1 17.9	17.7	15.7
Jewish	2.1	6.6	13.0	9.5	3.4 3.4	4.0	7.3
Oriental	1.0	1.2	3.0	2.9	4.1 5.0	1.4	2.4
None	23.0	4.9	16.3	14.1	11.4 22.4	30.1	18.5
Other	4.4	0.3	3.1	3.0	2.0 7.2	2.4	3.1 (62)

See Appendix 2.

A 3.9. Religious Practice of Physicians, by Province (%) (Q36)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Yes Occasionally	46.2 30.4	26.8 40.7	48.8 27.52		68.9 18.1	49.6 24.2	37.0 31.5	43.8 30.0
No	23.4	32.5	23.6	18.0	13.1	26.2	31.5	26.2 (190)

A 3.10. Ethnic Origin of Physicians,* by Province (%) (Q34)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
British	80.0	10.4	57.6	44.5	34.9	55.9	62.4	51.7
French	5.5	81.4	4.4	5.6	11.8	4.3	2.2	16.1
European								
(other/West)	7.9	-	13.8	30.5	18.1	15.0	14.7	12.5
European (East)	3.4	-	12.5	8.8	21.3	11.9	9.8	9.3
Jewish	1.7	7.0	3.6	2.7	-	2.1	2.5	3.3
Asian	1.4	1.2	8.1	7.8	13.9	10.8	8.4	7.1
								(149)

^{*} This question was not asked in the Quebec/France study. Results for Quebec are estimated based on questions about religion.

A 3.11. Place Where Physicians Received Their Medical Education, by Province (%) (Q16)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Alberta	2.3	-	3.6	4.2	4.2	54.6	13.8	11.2
British Columbia	2.2	-	0.9	-	2.9	3.8	35.5	8.8
Manitoba	0.2	0.9	2.1	56.4	3.3	1.1	3.7	4.7
Newfoundland	8.8	0.3	0.9	-	-	0.2	2.2	1.7
Nova Scotia	50.9	-	1.8	-	0.9	0.3	2.5	6.3
Ontario	11.3	2.0	63.1	6.9	3.6	10.7	10.5	26.4
Quebec	7.1	95.3	6.2	3.9	0.6	2.0	3.5	17.4
Saskatchewan	0.2	0.2	0.5	4.1	52.1	4.8	7.9	5.0
United States	2.1	0.1	1.7	0.8	3.3	0.8	1.6	1.4
Great Britain	10.7	0.2	10.0	13.3	11.9	15.7	13.3	10.3
European (other)	1.2	0.2	2.9	2.1	0.8	1.0	2.5	1.9
Africa	2.3	0.4	3.2	8.0	7.7	1.3	0.8	2.4
Asia	0.7	0.4	3.1	0.4	8.7	3.7	2.2	2.5

A 3.12. Practice Area of Physicians, by Province (%) (Q20)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Urban	69.2	83.3	73.5	65.7	63.7	70.6	70.6	72.8 27.2 (19)
Rural	30.8	16.7	26.5	34.3	36.3	24.9	29.4	

A 3.13. Socioeconomic Status of Physicians' Clientele, by Province (%) (Q26)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Well-to-do Middle-class Underprivileged Mixed	62.2 32.4 5.4	5.3 79.7 12.2 2.8	1.9 73.7 19.8 4.6	69.7 22.0 8.3	2.6 59.9 31.9 5.6	1.4 77.9 16.2 4.5	2.6 83.6 11.1 2.7	2.3 75.2 18.3 4.2 (49)

A 3.14. Type of Medical Practice of Physicians, by Province (%) (Q18)

ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
26.2	44.3	42.1	16.2	20.1	18.5	24.2	31.5
46.8	55.7	38.7	49.4	58.7	42.9	54.4	47.2
27.0	-	19.2	34.4	21.2	38.5	21.4	21.2
							(453)
16.5	30.6	18.3	25.3	10.3	16.1	6.1	17.2
19.2	34.4	21.5	13.7	29.0	32.2	27.1	25.6
1.1	2.6	1.8	2.9	3.5	0.6	0.2	1.5
63.2	32.5	58.4	58.0	57.2	51.1	66.6	55.7
	26.2 46.8 27.0 16.5 19.2	26.2 44.3 46.8 55.7 27.0 - 16.5 30.6 19.2 34.4 1.1 2.6	26.2 44.3 42.1 46.8 55.7 38.7 27.0 - 19.2 16.5 30.6 18.3 19.2 34.4 21.5 1.1 2.6 1.8	26.2 44.3 42.1 16.2 46.8 55.7 38.7 49.4 27.0 - 19.2 34.4 16.5 30.6 18.3 25.3 19.2 34.4 21.5 13.7 1.1 2.6 1.8 2.9	26.2 44.3 42.1 16.2 20.1 46.8 55.7 38.7 49.4 58.7 27.0 - 19.2 34.4 21.2 16.5 30.6 18.3 25.3 10.3 19.2 34.4 21.5 13.7 29.0 1.1 2.6 1.8 2.9 3.5	26.2 44.3 42.1 16.2 20.1 18.5 46.8 55.7 38.7 49.4 58.7 42.9 27.0 - 19.2 34.4 21.2 38.5 16.5 30.6 18.3 25.3 10.3 16.1 19.2 34.4 21.5 13.7 29.0 32.2 1.1 2.6 1.8 2.9 3.5 0.6	26.2 44.3 42.1 16.2 20.1 18.5 24.2 46.8 55.7 38.7 49.4 58.7 42.9 54.4 27.0 - 19.2 34.4 21.2 38.5 21.4 16.5 30.6 18.3 25.3 10.3 16.1 6.1 19.2 34.4 21.5 13.7 29.0 32.2 27.1 1.1 2.6 1.8 2.9 3.5 0.6 0.2

^{*} This question was asked differently in the Quebec/France survey. Multiplechoice question; percentages do not add up to 100%.

A 3.15. Distance of Physicians from a Genetics Centre,* by Province (%) (Q21)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Less than 100 km 100-250 km More than 250 km			78.1 14.8 7.1	24.9	33.5	64.3 22.3 13.4	64.5 10.2 25.3	67.6 17.3 15.0 (17)

^{*} This question was not asked in the Quebec/France survey. Distances are estimated.

A 3.16. Physicians' Source of Information on PND,* by Province (%) (Q27)

	ATL	ONT	MAN	SASK	ALB	BC/NWT	CAN
Conferences	27.2	33.4	29.8	56.2	30.0	28.8	32.0
Continuing education	56.6	52.0	57.1	55.4	56.7	68.4	57.8
Scientific journals	26.5	32.9	20.9	37.1	25.8	21.0	27.8
Colleagues	33.2	32.0	33.2	36.0	31.4	27.5	31.2
Medical press	41.6	48.1	37.0	45.5	40.6	38.8	43.2
Advertising	1.3	2.3	2.0	3.4	0.6	1.7	1.9
Other	0.3	1.8	4.9	0.8	2.5	2.4	2.0
							(486)

^{*} This question was not asked in the Quebec/France survey. Percentages do not add up to 100%.

A 3.17. Physicians Who Say They Are Directive in Advice to Patients,* by Province (%) (Q28)

	ATL	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not at all	25.5	28.7	28.8	12.4	26.3	20.7	25.1
Moderately	40.3	34.3	32.5	32.7	36.3	39.0	36.3
Totally	34.2	37.0	38.6	54.9	37.4	40.3	38.6 (518)

^{*} This question was not asked in the Quebec/France survey.

A 3.18. Physicians Who Say They Are Early Adopters of New Technology,* by Province (%) (Q28)

	ATL	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not at all Moderately Totally	38.2 35.6 26.1	36.6 35.6 27.8	32.0 36.7 31.3	33.3 28.2 38.5	29.5 40.4 30.1	30.7 36.6 32.7	33.9 36.2 29.9 (507)

^{*} This question was not asked in the Quebec/France survey.

A 3.19. Physicians Who Say They Consult Colleagues,* by Province (%) (Q28)

	ATL	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not at all	3.5	2.7	4.2	0.4	4.8	4.0	3.4
Moderately	9.7	14.9	14.5	14.0	10.0	10.4	12.4
Totally	86.8	82.5	81.3	85.7	85.2	85.6	84.2 (499)

^{*} This question was not asked in the Quebec/France survey.

A 3.20. Physicians Who Say They Discuss Opinions with Patients,* by Province (%) (Q28)

	ATL	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not at all	7.7	9.8	6.9	2.3	7.0	5.0	7.4
Moderately	18.0	16.0	14.4	16.6	12.5	14.4	15.3
Totally	74.3	74.1	78.6	81.1	80.5	80.6	77.3 (505)

^{*} This question was not asked in the Quebec/France survey.

A 3.21. Physicians Who Say They Are Conservative or Liberal,* by Province (%) (Q29)

	ATL	ONT	MAN	SASK	ALB	BC/NWT	CAN
Rather conservative Conservative and	32.3	33.9	30.4	37.4	46.0	30.0	34.5
liberal	36.7	37.2	36.1	44.6	35.3	39.0	37.7
Rather liberal	31.0	28.9	33.4	18.0	18.7	30.6	27.9 (500)

^{*} This question was not asked in the Quebec/France survey.

A 3.22. Average Number of Pregnancies Followed per Year, by Physician's Specialty and Province (Q23)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
GPs Number of	42.7	85.8	40.9	43.9	36.6	46.4	33.3	44.2
respondents Obstetricians Number of	(222) 194.0	(190) 199.4	(554) 287.0	(96) 360.4	(109) 251.1	(253) 256.9	(505) 271.0	255.5
respondents	(23)	(90)	(141)	(14)	(9)	(26)	(32)	

A 3.23. Average Number of Deliveries per Year, by Physician's Specialty and Province (Q23)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
GPs Number of	37.2	73.3	34.5	36.8	33.0	38.2	27.6	37.4
respondents Obstetricians	(219) 185.1	(186) 186.4	(545) 239.6	(96) 287.4	(101) 217.2	(250) 229.4	(491) 131.7	211.9
Number of respondents	(25)	(87)	(139)	(14)	(9)	(27)	(32)	

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Radiologists Number of	620.4	906.9	867.3	1 970.8	1 226.7 1	354.1	1 027.5	968.8
respondents	(25)	(40)	(71)	(6)	(8)	(29)	(39)	
Obstetricians Number of	196.4	555.5	381.4	299.5	306.5	480.3	253.8	401.6
respondents	(12)	(42)	(81)	(7)	(8)	(17)	(15)	
GPs Number of	43.6	105.9	46.1	47.6	46.8	42.8	37.7	47.8
respondents	(163)	(110)	(413)	(64)	(90)	(184)	(396)	

A 3.25. Average Number of Newborns Taken Under Care per Year, by Physician's Specialty and Province (Q23)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Paediatricians Number of	270.7	262.6	218.4	134	349.5	335	252	245.6
respondents GPs	(21) 41	(118) 60.4	(143) 39	(22) 37.1	(8) 34.2	(30) 44.6	(35) 29.9	39.1
Number of respondents	(215)	(169)	(548)	(96)	(105)	(253)	(500)	

A 3.26. Average Number of Amniocenteses Ordered per Year, by Physician's Specialty and Province (%) (Q25)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
GPs Number of	2.6	4.6	6.9	2.9	2.7	5.9	6	5.4
respondents Obstetricians Number of	(126) 11.0	(159) 15.7	(369) 19.6	(69) 24.3	(56) 22.4	(158) 22.9	(384) 17.6	18.3
respondents	(23)	(87)	(132)	(13)	(9)	(27)	(31)	

A 3.27. Average Number of CVSs Ordered per Year, by Physician's Specialty and Province (Q25)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
GPs Number of	1.2	1.5	1.7	2.5	1.0	2.5	1.9	1.9
respondents Obstetricians Number of	(19) 2.5	,	(149) 7.2	(38) 11.1	(11) 1.5	(62) 11.1	(212) 10.5	7.4
respondents	(7)	(41)	(104)	(12)	(1)	(22)	(28)	

A 3.28. Average Number of Serum Alpha-Fetoprotein Tests Ordered per Year, by Physician's Specialty and Province (Q25)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
GPs Number of	2.4	4.6	18.3	32.2	2.0	5.7	2.9	10.9
respondents Obstetricians Number of	(98) 17.1	, ,	(388) 108.9	(87) 307.3	,	(118) 17.2	(294) 22.1	71.8
respondents	(17)	(64)	(120)	(11)	(7)	(22)	(26)	

A 3.29. Average Percentage of Pregnant Women Receiving One Ultrasound Scan per Year, by Physician's Specialty and Province (Q25)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
GPs Number of	91.2	94.2	88.1	69.4	90.0	79.0	90.7	87.8
respondents Obstetricians Number of	(217) 87.8	(190) 97.4	(548) 92.4	(96) 61.5	(105) 90.8	(246) 87.1	(510) 92.4	91.6
respondents	(3)	(89)	(140)	(14)	(10)	(28)	(33)	

A 3.30. Physicians Who Ordered Ultrasound Scans for Their Patients, by Specialty and Province (%) (Q24)

	ATL	QUE	ONT	MAN.	SASK	ALB	BC/NWT	CAN
Obstetrician	0.5	21.4	6.3	4.5	-	1.4	4.4	6.1
Radiologist Obstetrician or	80.3	67.2	70.9	60.7	82.2	80.6	85.9	76.3
radiologist	17.1	11.4	12.2	26.4	16.9	9.7	5.3	11.6
Other	2.1	-	10.5	8.4	0.8	8.3	4.4	6.0 (51)

A 3.31. Physicians and Reaction to Questionnaire, by Province (%) (Q37)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Totally positive								
Interest	85.2	91.7	82.6	80.6	80.5	83.6	80.5	83.7
Clarity	66.1	80.8	72.2	71.8	71.9	67.5	65.1	70.8
Relevance	71.2	81.4	72.3	72.6	74.1	70.9	71.1	73.3
Length	41.1	50.6	46.4	37.8	51.6	44.2	44.5	45.7

Chapter 4. Frequency Tables by Province

A 4.1. Number of Ultrasound Scans Considered Appropriate in Course of Normal Pregnancy (%) (Q1)

n	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
	304	475	999	152	144	353	645	3 072
0 ultrasound 1 2 3	18.9 75.0 5.8 0.3	4.3 55.2 38.2 2.2	20.4 63.9 13.8 1.9	43.0 50.1 6.1 0.8	10.5 59.6 26.0 3.9	36.6 57.0 5.6 0.8	17.1 68.6 13.8 0.5	19.6 63.0 16.0 1.4 (64)*

 $p \le 0.01$

A 4.2. Acceptability of Reasons for Using Ultrasound Scanning (%) (Q3)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Obstetrical data								
Not justified (1-2)	5.0	3.2	5.0	6.5	3.4	6.9	7.2	5.4
Moderately justified (3)	7.2	6.8	9.4	11.4	8.0	9.1	7.5	8.4
Totally justified (4-5)	87.8	90.0	85.6	82.1	88.6	84.0	85.3	86.2
								(48)
								` /
Screening for								
malformations								
Not justified	18.6	5.3	22.6	30.7		25.2	25.8	21.4
Moderately justified	21.8	5.7	19.3	24.4	21.2	25.0	14.5	17.4
Totally justified	59.6	89.0	58.1	44.9	45.2	49.8	59.7	61.1
								(63)
December weren								
Reassuring women	05.0	00.0	00.4	40.0		07.0		
Not justified	35.6	26.9	33.4	42.9	44.1	27.6	36.3	33.5
Moderately justified	37.1	39.3	37.2	30.0	2 5.9	41.1	42.0	38.1
Totally justified	27.4	33.8	29.4	27.0	30.0	31.2	21.8	28.4
								(72)

^{*} The number of non-respondents appearing in parentheses applies to Canada as a whole.

(48)

54.3

30.5

15.2 (68)

42.2

30.6

27.1

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Giving sense of responsibility								
Not justified	71.1	58.7	68.9	67.9	59.2	63.6	62.5	65.1
Moderately justified	18.9	25.8	19.6	27.9	28.2	25.6	23.5	22.8
Totally justified	9.4	15.5	11.5	4.2	12.6	10.8	14.1	12.0 (47)
Enabling parents to view fetus								
Not justified	78.3	67.4	78.0	88.2	68.4	81.9	78.8	77.1
Moderately justified	17.9	24.0	15.6	8.8	21.1	13.6	15.8	16.8
Totally justified	3.8	8.7	6.4	3.0	10.4	4.6	5.4	6.1 (48)
Learn sex of fetus								
Not justified	99.0	94.7	97.7	98.7	94.0	99.5	98.0	97.5
Moderately justified	0.9	3.9	1.5	1.3	6.0	0.3	1.0	1.8
Totally justified	0.2	1.4	0.8	0.0	0.0	0.2	0.9	0.7

57.1

13.4

56.0

15.0 **11.9**

29.5 29.0

59.2

28.9 26.7

62.8

10.5

60.4 52.0

34.4 37.2

5.2 10.8

 $p \le 0.01$

Professional use Not justified

Moderately justified

Totally justified

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Woman decides	68.6	71.3	69.6	62.7	77.9	66.1	75.7	70.7
Exam not important Do not accept refusal and suggest seeing	30.8	19.5	28.8	36.9	21.7	33.8	20.8	26.5
another doctor	0.6	9.1	1.6	0.4	0.4	0.2	3.5	2.8 (86)

A 4.4. Ultrasound Should Be Subject to Written Prior Agreement from Patient (%) (Q15 #13)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree	49.8	64.7	59.6	48.8	64.8	57.8	58.8	58.7
Moderately agree	25.0	12.9	16.5	27.4	10.8	19.3	12.4	16.5
Totally agree	25.5	22.4	23.9	23.8	24.4	22.9	28.8	24.7 (31)

 $p \le 0.01$

A 4.5. Perception of Reliability of Obstetrical Ultrasound in Detecting Malformations at 16-20 Weeks' Gestation (%) (Q4)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Anencephaly								
Not reassured	3.9	3.7	4.0	8.9	17.4	5.2	3.6	4.9
Moderately reassured	6.3	4.8	7.6	6.6	15.1	5.2	7.3	7.0
Totally reassured	89.8	91.5	88.4	84.5	67.6	89.7	89.0	88.1 (54)
Spina bifida								
Not reassured	21.3	19.1	25.4	27.5	39.3	30.7	26.8	25.7
Moderately reassured	33.5	35.9	35.8	33.9	30.9	30.2	39.9	35.5
Totally reassured	45.2	45.0	38.8	38.6	29.8	39.0	33.2	38.8
								(58)
Limb malformation								
Not reassured	36.4	26.2	28.8	23.5	47.6	24.7	26.7	28.9
Moderately reassured	28.0	26.6	29.9	31.2	27.2	34.5	28.0	29.3
Totally reassured	35.6	47.3	41.3	45.5	25.2	40.8	45.3	41.9
								(62)
Hydrocephaly								
Not reassured	20.1	24.3	24.7	34.4	29.6	33.7	31.0	27.3
Moderately reassured	34.8	29.1	27.1	20.8	36.0	24.4	27.6	27.8
Totally reassured	45.1	46.6	48.2	44.8	34.5	44.0	41.4	44.9
·								(60)
Heart malformation								
Not reassured	57.5	60.5	54.0	56.5	60.9	53.1	49.2	57.4
Moderately reassured	27.6	28.7	33.9	31.8	26.2	27.6	32.2	30.0
Totally reassured	15.0	10.9	12.1	11.6	12.9	19.3	18.6	14.4
•								(64)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Trisomy 21 (without structural malformations)								
Not reassured '	85.7	93.6	86.0	93.7	71.2	82.1	91.2	87.4
Moderately reassured	11.6	4.4	9.9	5.2	16.9	14.3	7.8	9.4
Totally reassured	2.7	2.0	4.1	1.1	11.9	3.6	1.0	3.2
•								(63)

A 4.6. Age at Which Amniocentesis Should Be Available to Women, Irrespective of Present Policies (%) (Q5A)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
≤34 years	4.9	10.6	8.6	10.3	6.4	7.6	14.9	9.7
35 years	65.6	66.8	62.2	64.4	44.1	64.6	58.9	62.1
36-39 years	10.7	15.7	16.5	13.3	30.4	12.6	15.8	15.7
≥40 years	12.5	3.7	7.7	3.8	5.4	13.0	4.7	7.2
Never	6.3	3.2	5.0	8.3	13.7	2.2	5.7	5.2
								(74)

 $p \le 0.01$

A 4.7. Age at Which CVS Should Be Available to Women, Irrespective of Present Policies (%) (Q5B)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
≤34 years	5.1	8.8	5.7	9.7	6.0	6.9	14.0	8.3
35 years	52.5	42.2	53.6	56.8	35.8	55.1	54.2	51.4
36-39 years	12.9	11.3	18.9	14.3	25.3	8.6	17.4	15.7
≥40 years	17.1	15.0	10.4	3.4	6.4	16.2	6.9	11.1
Never	12.4	22.7	11.4	15.7	26.5	13.2	7.6	13.5
								(31)

A 4.8.	Acceptability o	f Rea	sons	for Us	sing A	mnioce	entesi	s (%)
	A ⁻	rL C	QUE	ONT	MAN	SASK	ALB	BC/NW

Anxiety Not justified Moderately justified Totally justified Freedom of choice (age 33) Not justified Moderately justified Totally justified Access without criteria	59.4 25.9 14.7 57.4 23.5 19.1	43.8 25.7 30.5 48.5 22.1 29.3	55.3 22.9 21.8 57.9 20.2 21.9	46.2 26.7 27.0 49.6 22.7 27.7	49.3 28.1 22.6 51.6 26.9 21.5	59.5 24.3 16.2 59.2 19.6 21.3	59.8 20.2 20.0 65.2 18.2	54.6 23.7 21.7 (37)
Moderately justified Totally justified Freedom of choice (age 33) Not justified Moderately justified Totally justified Access without	25.9 14.7 57.4 23.5	25.7 30.5 48.5 22.1	22.9 21.8 57.9 20.2	26.7 27.0 49.6 22.7	28.1 22.6 51.6 26.9	24.3 16.2 59.2 19.6	20.2 20.0 65.2	23.7 21.7 (37)
Totally justified Freedom of choice (age 33) Not justified Moderately justified Totally justified Access without	14.7 57.4 23.5	48.5 22.1	21.8 57.9 20.2	27.0 49.6 22.7	22.6 51.6 26.9	16.2 59.2 19.6	20.0 65.2	21.7 (37)
Freedom of choice (age 33) Not justified Moderately justified Totally justified Access without	57.4 23.5	48.5 22.1	57.9 20.2	49.6 22.7	51.6 26.9	59.2 19.6	65.2	(37) 57.
(age 33) Not justified Moderately justified Totally justified Access without	23.5	22.1	20.2	22.7	26.9	19.6		57.4
(age 33) Not justified Moderately justified Totally justified Access without	23.5	22.1	20.2	22.7	26.9	19.6		
Not justified Moderately justified Totally justified Access without	23.5	22.1	20.2	22.7	26.9	19.6		
Moderately justified Totally justified Access without	23.5	22.1	20.2	22.7	26.9	19.6		
Totally justified Access without							18.2	
Access without	19.1	29.3	21.9	27.7	21.5	27 2	40.0	
						21.3	16.6	21. 9 (51)
criteria								, ,
(against payment) (Q15 #5)								
Not justified	37.7	37.4	24.7	25.8	34.1	26.5	18.4	7.
Moderately justified	13.1	15.0	14.1	15.6	19.4	16.1	9.1	13.
Totally justified	49.2	47.5	61.2	58.6	46.4	57.4	72.4	59.0
								(15)
Access without								
criteria								
(public health								
system) (Q15 #27)		04.0	70.0	70.0	07.4	70.0	04.77	
Not justified	80.5	61.6	76.8	78.0	67.1	76.0	81.7	75.4
Moderately justified	12.8	12.3	9.9	9.7	19.7	8.7	7.6	10.4
Totally justified	6.7	26.1	13.3	12.3	13.2	15.3	10.7	14. (32)
Learning sex of								,
fetus (CVS)								
Not justified	92.1	89.3	93.5	94.0	93.2	94.7	88.7	91.8
Moderately justified	0.6	4.6	4.2	3.3	3.7	3.4	7.6	4.
Totally justified	7.3	6.1	2.3	2.7	3.0	1.9	3.8	3.
, ,								(47)
If abortion is								
refused			40.0					
Not justified	57.5	52.5	48.9	30.5	55.4	50.8	53.2	50.
Moderately justified	14.5	11.7	14.7	16.9	18.6	14.1	8.0	13.0
Totally justified	28.0	35.8	36.4	52.6	26.0	35.2	38.8	36. 2 (42)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
36-year-old woman								
Recommends procedure Does not recommend	36.4	67.2	38.6	41.5	36.1	41.3	49.2	45.4
procedure	20.7	9.0	12.7	14.0	16.1	13.8	22.8	15.4
Recommends a screening	3							
ultrasound	18.5	17.9	21.6	15.3	35.6	21.1	10.0	18.6
Other	28.5	6.9	33.2	32.6	14.1	29.4	22.8	25.1 (58)
38-year-old woman								
Recommends procedure	53.5	83.1	56.0	56.1	52.5	61.1	66.8	62.6
Does not recommend								
procedure	10.7	3.8	6.9	5.6	10.1	6.9	10.3	7.6
Recommends a screening)							
ultrasound	14.7	8.8	14.8	11.9	27.3	11.4	4.9	11.8
Other	24.7	5.4	27.2	31.0	10.8	25.0	20.0	21.2 (51)

A 4.10. Fear of Lawsuits Makes Us Use PND More Often Than Would Be Medically Indicated (%) (Q15 #32)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	21.5 16.6 61.9	41.6 18.8 39.6	24.5 17.1 58.3	28.0 15.1 56.9	12.1 22.7 65.2	25.6 15.1 59.3	32.7 10.7 56.6	28.3 15.9 55.8 (50)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Diabetes								
In utero	6.4	11.2	7.7	7.3	7.5	4.4	6.7	7.5
Childhood	58.7	68.2	63.4	62.2	74.8	67.7	73.2	66.7
Adulthood	21.9	8.9	15.7	20.7	10.7	19.6	14.4	15.5
Never	13.0	11.7	13.2	9.8	7.0	8.3	5.7	10.3
								(18)
Alcoholism								
In utero	2.8	6.0	4.2	1.9	3.4	4.1	3.4	4.0
Childhood	46.2	49.6	44.3	51.0	58.2	50.4	57.5	49.7
Adulthood	36.6	27.3	33.6	29.6	31.7	30.9	28.0	31.2
Never	14.5	17.1	17.9	17.6	6.7	14.6	11.1	15.1
								(24)
Schizophrenia								
In utero	12.3	27.1	16.3	4.2	10.6	15.9	19.9	17.4
Childhood	35.9	42.3	42.0	58.2	55.2	45.3	44.4	43.7
Adulthood	22.2	11.0	18.7	13.0	27.3	20.2	16.1	17.6
Never	29.5	19.7	23.0	24.5	6.9	18.5	19.7	21.3
								(34)
Alzheimer's disease								
In utero	4.0	14.1	8.7	1.9	4.4	8.3	8.9	8.5
Childhood	15.1	16.7	17.7	26.0	25.3	20.1	17.7	18.3
Adulthood	45.2	41.8	40.6	39.4	52.1	43.7	42.4	42.4
Never	35.7	27.5	33.1	32.7	18.2	27.9	31.0	30.7
								(33)
Coronary heart								
disease								
In utero	2.6	5.4	3.7	2.4	3.0	3.3	2.4	3.4
Childhood	53.1	53.3	52.6	52.6	48.7	56.6	60.8	54.8
Adulthood	34.9	31.9	34.4	36.8	42.5	34.2	32.4	34.1
Never	9.4	9.4	9.3	8.2	5.7	5.9	4.4	7.7
								(16)

A 4.12. Acceptability of Reasons for Using Predisposition Testing (%) (Q12B)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Diabetes								
To prevent births	.1.5	4.3	3.0		3.9	1.4	4.4	3.1
Early treatment	55.6	50.5	57.0	62.3	76.6	64.0	67.5	60.1
Preventive counselling	33.4	37.1	31.0	30.3	10.0	29.2	23.5	29.4
None	9.5	8.2	9.0	7.3	9.4	5.3	4.6	7.5
								(26)
Alcoholism								
To prevent births	1.3	2.9	2.0	0.4	0.7	0.5	1.8	1.7
Early treatment	14.9	15.0	16.5	16.3	20.4	22.9	17.1	17.2
Preventive counselling	75.8	71.6	70.7	75.0	72.1	68.0	74.3	72.1
None	8.0	10.5	10.8	8.4	6.7	8.6	6.8	9.1
								(20)
Sobizophronia								
Schizophrenia To prevent births	9.8	26.9	14.9	4.8	7.7	13.8	20.6	16.5
Early treatment	48.4	32.1	45.7	55.2	62.3	54.5	44.9	46.4
Preventive counselling	16.3	19.3	18.8	17.0	17.6	16.9	16.6	17.8
None None	25.5	21.7	19.3	23.0	12.4	14.8	17.9	19.3
None	25.5	21.7	19.3	23.0	12.4	14.0	17.9	(37)
								(07)
Alzheimer's disease								
To prevent births	2.7	11.3	7.0	1.2	1.7	7.5	8.0	7.0
Early treatment	33.6	24.2	29.1	29.9	40.5	36.2	28.6	30.1
Preventive counselling	24.9	29.3	30.7	36.3	37.1	28.6	30.0	30.1
None	38.8	35.2	33.1	32.6	20.7	27.7	33.4	32.8
								(54)
Coronary heart								
disease								
To prevent births	1.1	1.3	0.8	-	-	0.6	1.0	0.9
Early treatment	38.3	26.3	33.2	36.6	43.8	38.2	37.7	34.9
Preventive counselling	55.2	65.9	59.9	59.3	48.0	56.8	59.6	59.3
None	5.4	6.5	6.0	4.0	8.2	4.4	1.7	4.9
								(18)

A 4.13. Predisposition Testing in Absence of Available Treatment Useless (%) (Q15 #15)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	48.5 22.3 29.2	45.0 17.3 37.7	49.6 15.6 34.8	54.3 15.9 29.8	46.8 19.8 33.4	49.4 18.7 32.0	56.6 14.4 29.0	50.3 16.8 32.8
Totally agree	29.2	37.7	34.0	29.0	33.4	32.0	29.0	(38)

A 4.14. It is Acceptable to Recommend Surrogate Motherhood to Couples When Female Partner Has Dominant Genetic Disorder (%) (Q15 #16)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not acceptable Moderately acceptable Totally acceptable	37.1	27.5	28.5	28.4		28.5	31.2 27.9 40.9	30.7 28.9 40.4 (68)

 $p \le 0.01$

A 4.15. It is Acceptable to Recommend Artificial Insemination to Couples When Male Partner Has Dominant Genetic Disorder (%) (Q15 #22)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not acceptable Moderately acceptable Totally acceptable	23.2	19.7	14.0	12.2	23.3	16.3	7.9 14.2 77.9	9.2 16.4 74.3 (32)

A 4.16. Predetermining an Embryo's Sex by Chromosome Selection (%) (Q15 #9)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not acceptable Moderately acceptable Totally acceptable	18.4	20.1	11.6	4.5		84.3 6.8 9.0	71.4 12.1 16.6	72.3 12.9 14.9 (33)

 $p \le 0.01$

A 4.17. Authorizing Marketing of Self-Prescribed Tests to Determine Sex of Fetus (%) (Q15 #25)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not acceptable Moderately acceptable Totally acceptable	22.8	13.3	19.4	19.5			67.9 16.4 15.7	69.0 17.5 13.5 (23)

A 4.18. Perception of Parents' Difficulties When Offspring Have Various Conditions (%) (Q9A, Q10)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Aggressiveness								
Not difficult	12.4	9.8	13.1	12.7	12.5	10.9	10.6	11.7
Moderately difficult	34.5	34.3	31.2	32.5	51.1	31.7	30.6	32.9
Totally difficult	53.1	55.9	55.7	54.8	36.5	58.0	58.8	55.4
•								(28)
Behavioural								
problems								
Not difficult	7.4	7.9	9.2	7.0	11.2	6.6	4.2	7.4
Moderately difficult	32.1	28.5	31.3	33.4	29.4	29.0	28.8	30.2
Totally difficult	60.5	63.6	59.5	59.6	59.4	64.4	67.1	62.4
								(21)
Learning disabilities								
Not difficult	17.6	8.7	21.1	14.5	14.2	14.2	15.8	16.3
Moderately difficult	40.9	35.1	36.2	38.4	41.0	41.6	39.9	38.2
Totally difficult	41.5	56.2	42.7	47.1	44.8	44.3	44.3	45.5
								(31)
Female sterility								
Not difficult	67.4	42.6	61.6	55.4	55.6	59.7	63.6	58.8
Moderately difficult	20.3	35.0	23.5	29.1	28.5	26.6	24.6	26.
Totally difficult	12.3	22.4	14.9	15.5	16.0	13.7	11.9	15.
								(11)
Male sterility								
Not difficult	67.3	43.9	64.4	57.4	58.2	61.5	66.8	61.
Moderately difficult	23.9	34.6	22.8	28.7	26.7	27.5	22.4	25.
Totally difficult	8.8	21.6	12.8	13.9	15.2	10.9	10.7	13.
								(8)
Hypogonadism								
Not difficult	50.3	30.3	47.8	47.5	50.0	53.8	48.5	46.
Moderately difficult	27.9	39.7	32.2	30.1	18.1	30.5	33.2	32.
Totally difficult	21.8	30.1	19.9	22.5	31.8	15.6	18.3	21.
								(27)
XXX syndrome								
Not severe	39.3	31.8	39.8	38.5	32.4	38.6	35.6	37.
Moderately severe	39.6	38.9	36.5	40.0	45.4	41.2	41.4	39.
Very severe	21.1	29.3	23.7	21.5	22.2	20.2	22.9	23.
								(65)

A 4.18. (cont'd)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Klinefelter's								
syndrome								
Not severe	29.5	24.9	33.5	39.8	39.4	32.7	32.0	31.9
Moderately severe	42.9	41.3	40.2	40.5	38.1	41.7	38.6	40.4
Very severe	27.7	33.9	26.3	19.7	22.6	25.7	29.4	27.7
,						20.7	20.4	(60)
XYY syndrome								
Not severe	42.7	38.4	43.5	41.7	37.5	41.2	36.1	40.4
Moderately severe	36.4	36.1	36.0	38.3	37.8	40.5	41.4	37.9
Very severe	20.9	25.4	20.5	20.0	24.7	18.4	22.5	21.7
,				20.0		10.4	22.0	(66)
Severe cleft lip and								
palate								
Not difficult	29.6	12.0	26.4	23.8	18.7	28.4	25.7	24.1
Moderately difficult	24.9	27.3	25.3	35.1	21.4	29.6	28.8	27.1
Very difficult	45.5	60.7	48.2	41.1	59.9	42.0	45.5	48.8
•								(16)
Lobster claw								
deformity								
Not difficult	31.2	14.4	33.2	31.0	29.0	32.5	30.7	29.2
Moderately difficult	30.1	27.3	28.5	30.8	17.9	32.4	34.1	29.7
Very difficult	38.7	58.3	38.3	38.2	53.1	35.1	35.2	41.1
•								(13)
Intellectual								
impairment								
Not difficult	19.2	4.9	17.1	7.9	15.4	16.5	10.4	13.4
Moderately difficult	31.2	10.8	26.6	30.7	28.0	31.5	26.8	25.5
Very difficult	49.6	84.3	56.3	61.5	56.6	52.0	62.8	61.1
								(20)
Paraplegia								
Not difficult	8.7	2.0	5.2	4.3	7.2	3.3	6.9	5.3
Moderately difficult	13.4	3.4	12.5	13.9	9.3	15.7	10.7	11.1
Very difficult	77.9	94.6	82.3	81.8	83.5	81.0	82.4	83.7
•								(18)

A 4.19. I Could Not Accept the Idea of Having a Child with Trisomy 21 (%) (Q15 #30)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	47.7 20.0 32.3	20.4 13.9 65.8		23.4	47.4 35.6 16.9	47.6 17.8 34.6	42.4 16.7 40.9	40.1 19.8 40.2 (57)

A 4.20.	Acceptability	of Abortion	for Certain	Conditions	(%) (Q11)
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ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
37.9	17.2	34.3	40.7	55.6	42.3	26.8	32.7
13.3	12.3	18.1	20.8	19.8	9.9	19.2	16.2
48.8	70.4	47.6	38.5	24.6	47.8	54.0	51.1 (37)
40.1	24.7	31.0	42.9	56.5	36.0	27.0	32.5
12.8	16.7	15.3	15.7	22.7	12.9	19.2	16.2
47.1	58.6	53.7	41.4	20.8	51.1	53.7	51.3 (27)
38.7	24.0	32.9	39.1	58.3	41.2	30.4	34.1
10.5	19.4	15.6	17.2	12.7	14.6	11.6	14.7
50.9	56.5	51.4	43.7	28.9	44.2	57.9	51.3 (34)
45.9	27.1	37.5	41.7	52.4	45.6	33.3	37.7
19.2	18.8	20.1	22.7	19.9	14.1	19.2	19.1
34.8	54.1	42.4	35.6	27.7	40.3	47.4	43.2 (58)
	37.9 13.3 48.8 40.1 12.8 47.1 38.7 10.5 50.9	37.9 17.2 13.3 12.3 48.8 70.4 40.1 24.7 12.8 16.7 47.1 58.6 38.7 24.0 10.5 19.4 50.9 56.5 45.9 27.1 19.2 18.8	37.9 17.2 34.3 13.3 12.3 18.1 48.8 70.4 47.6 40.1 24.7 31.0 12.8 16.7 15.3 47.1 58.6 53.7 38.7 24.0 32.9 10.5 19.4 15.6 50.9 56.5 51.4 45.9 27.1 37.5 19.2 18.8 20.1	37.9 17.2 34.3 40.7 13.3 12.3 18.1 20.8 48.8 70.4 47.6 38.5 40.1 24.7 31.0 42.9 12.8 16.7 15.3 15.7 47.1 58.6 53.7 41.4 38.7 24.0 32.9 39.1 10.5 19.4 15.6 17.2 50.9 56.5 51.4 43.7	37.9 17.2 34.3 40.7 55.6 13.3 12.3 18.1 20.8 19.8 48.8 70.4 47.6 38.5 24.6 40.1 24.7 31.0 42.9 56.5 12.8 16.7 15.3 15.7 22.7 47.1 58.6 53.7 41.4 20.8 38.7 24.0 32.9 39.1 58.3 10.5 19.4 15.6 17.2 12.7 50.9 56.5 51.4 43.7 28.9 45.9 27.1 37.5 41.7 52.4 19.2 18.8 20.1 22.7 19.9	37.9 17.2 34.3 40.7 55.6 42.3 13.3 12.3 18.1 20.8 19.8 9.9 48.8 70.4 47.6 38.5 24.6 47.8 40.1 24.7 31.0 42.9 56.5 36.0 12.8 16.7 15.3 15.7 22.7 12.9 47.1 58.6 53.7 41.4 20.8 51.1 38.7 24.0 32.9 39.1 58.3 41.2 10.5 19.4 15.6 17.2 12.7 14.6 50.9 56.5 51.4 43.7 28.9 44.2 45.9 27.1 37.5 41.7 52.4 45.6 19.2 18.8 20.1 22.7 19.9 14.1	37.9 17.2 34.3 40.7 55.6 42.3 26.8 13.3 12.3 18.1 20.8 19.8 9.9 19.2 48.8 70.4 47.6 38.5 24.6 47.8 54.0 40.1 24.7 31.0 42.9 56.5 36.0 27.0 12.8 16.7 15.3 15.7 22.7 12.9 19.2 47.1 58.6 53.7 41.4 20.8 51.1 53.7 38.7 24.0 32.9 39.1 58.3 41.2 30.4 10.5 19.4 15.6 17.2 12.7 14.6 11.6 50.9 56.5 51.4 43.7 28.9 44.2 57.9 45.9 27.1 37.5 41.7 52.4 45.6 33.3 19.2 18.8 20.1 22.7 19.9 14.1 19.2

A 4.20. (cont'd)

	ATI	OUE	ONIT					
	ATL	QUE	ONT	MAN	SASI	ALB	BC/NWT	CAN
Cystic fibrosis								
Not acceptable	52.8	32.1	42.4	48.9	71.4	47.8	41.6	44.0
Moderately acceptable	16.0	20.9	18.6	24.3	15.3	15.9	19.3	18.6
Totally acceptable	31.2	47.0	39.0	26.8	13.3	36.3	39.1	37.4
								(23)
Spina bifida								
Not acceptable	49.7	38.6	47.6	56.7	63.3	56.4	43.9	47.8
Moderately acceptable	25.7	23.4	20.8	21.4		17.8	26.7	22.8
Totally acceptable	24.5	38.0	31.7	22.0	10.9		29.4	
Totally acceptable	24.0	50.0	51.7	22.0	10.9	25.6	29.4	29.3 (27)
Dhanyllatan								(=1)
Phenylketonuria	67.1	40.0	GE O	77.4	70.4	05.0	50.0	
Not acceptable	67.1	49.8	65.2	77.1	79.4		58.6	62.9
Moderately acceptable	16.1	20.6	16.0	11.3	12.7		15.0	15.5
Totally acceptable	16.8	29.6	18.8	11.6	7.9	23.9	26.4	21.6
								(43)
Turner's syndrome								
Not acceptable	63.0	48.2	62.6	68.5	69.2	65.8	53.9	59.6
Moderately acceptable	12.5	24.6	19.0	14.6	20.1	16.9	21.9	19.4
Totally acceptable	24.5	27.2	18.4	16.9	10.7	17.3	24.2	21.0
								(46)
Klinefelter's syndrome								
Not acceptable	66.9	53.4	65.1	66.1	78.9	66.6	59.9	63.2
Moderately acceptable	17.6	24.9	17.7	19.8		18.9	20.6	19.5
Totally acceptable	15.5	21.7	17.2	14.1		14.5	19.5	17.2
rotany acceptable	10.0	21.7	17.5	17.1	5.1	14.5		(27)
								(21)
XYY syndrome								
Not acceptable	66.8	56.5	65.4	67.0	79.0	66.6	60.4	64.0
Moderately acceptable	17.3	23.8	19.1	20.1	15.3	18.1	20.7	19.7
Totally acceptable	16.0	19.7	15.5	12.8	5.7	15.3	19.0	16.3
								(31)
XXX syndrome								
Not acceptable	68.3	55.2	65.9	65.5	78.9	67.7	62.6	64.6
Moderately acceptable	18.7	24.7	18.3	21.4		19.0	19.0	19.5
Totally acceptable	13.0	20.1	15.9	13.1		13.3	18.4	15.9
otan, doooptable	.0.0	20.7	. 0.0	.0.1	0.1	10.0		(24)
								(24)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Lobster claw deformity								
Not acceptable	78.6	63.8	74.5	77.4	82.9	79.7	70.4	73.6
Moderately acceptable	12.3	20.9	14.9	16.3	13.7	13.1	17.9	16.0
Totally acceptable	9.1	15.3	10.5	6.4	3.4	7.2	11.7	10. 4 (29)
Fetus of non-desired sex								
Not acceptable	97.2	96.5	98	98.4	99.7	98.6	96.5	97.5
Moderately acceptable	0.3	0.8	1.0	1.6	-	0.3	1.7	1.0
Totally acceptable	2.4	2.7	1.1	-	0.3	1.1	1.7	1. (13)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree	33.1	31.9	33.3	29.6	21.8	31.1	31.7	31.8
Moderately agree	14.6	15.4	11.9	12.0	13.9	10.2	8.5	11.9
Totally agree	52.3	52.6	54.9	58.4	64.3	58.7	59.8	56.4 (59)

A 4.22. Aborting a Fetus with an Anomaly Is More Justifiable in the
First Than in the Second Trimester of Pregnancy (%) (Q15 #21)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	34.4 14.2 51.5	40.2 7.7 52.2	38.1 13.5 48.4	38.9 15.8 45.3	48.4 16.7 34.9	14.3	35.9 15.8 48.3	39.7 13.5 46.7 (49)

A 4.23. Aborting a Fetus with a Minor Anomaly Is Justifiable (%) (Q15 #12)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	69.3 14.8 16.0	17.6	69.1 15.4 15.5	72.5 15.3 12.2	86.9 8.5 4.7	73.0 13.1 14.0	67.7 16.4 15.9	70.6 15.3 14.1 (29)

 $p \le 0.01$

A 4.24. One Must Condemn PND Done with the Deliberate Intention of Terminating the Pregnancy if the Results Reveal the Existence of Anomaly (%) (Q15 #10)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	64.6 15.8 19.6	78.3 9.3 12.5	71.3 10.2 18.5		53.8 12.9 33.4	68.0 11.0 21.0	78.9 6.3	72.0 10.0 18.0
						2110	1 1.0	(39)

 $p \le 0.01$

A 4.25. It is Acceptable to Encourage Women with an Anencephalic Fetus to Continue Their Pregnancy so That the Fetus's Healthy Organs Can Be Used for Transplants (%) (Q15 #29)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	51.9 25.0 23.1	68.0 15.2 16.8	48.4 23.1 28.6	40.4 30.8 28.8	50.6 20.2 29.2	58.4 20.6 21.0	48.4 27.0 24.6	52.6 22.8 24.6 (59)

A 4.26. A Physician Must Be Able to Resist Some Abortion Requests When of the Opinion the Anomaly Is Minor (%) (Q15 #2)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	29.5 8.4 62.1	23.7 17.0 59.3	24.9 13.5 61.6	21.8 5.1 73.1	14.6 9.3 76.2	18.7 16.4 64.9	26.4 13.0 60.6	24.1 13.1 62.7 (30)

A 4.27. Physicians, Not Parents, Should Decide Which Fetal Anomalies Warrant Pregnancy Termination (%) (Q15 #6)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	59.2 22.8 18.0	46.9 28.5 24.6	64.4 22.5 13.1	56.3 25.1 18.6	54.4 30.9 14.7	60.7 25.7 13.6	62.4 25.1 12.5	59.5 24.9 15.6 (30)

 $p \le 0.01$

A 4.28. Parents Have an Absolute Right to Freedom of Choice with Respect to Abortion (%) (Q15 #4)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	43.0 13.9 43.1	27.0 15.1 57.9	31.8 15.3 52.8	45.6 14.9 39.5	55.4 9.4 35.1	47.0 13.0 40.0	32.0 15.5 52.4	35.8 14.6 49.6 (17)

A 4.29. A Physician Must Discuss the Question of Abortion with Alcoholic Women (%) (Q15 #24)

-	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	43.9 30.3 25.8	36.5 28.8 34.7	41.0 30.6 28.4	52.3 22.9 24.7	56.0 22.3 21.7	44.7 26.5 28.8	40.0 21.0 39.1	42.1 27.0 30.9 (36)

A 4.30. Should Parents Be Told if the Fetus Carries a Sex Chromosome Anomaly? (%) (Q9B)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
XYY syndrome								
Yes	92.3	94.2	93.5	92.9	89.2	95.7	97.7	94.4
No	7.7	5.8	6.5	7.1	10.8	4.3	2.3	5.6
XXY syndrome								
Yes	98.2	98.5	97.3	97.6	92.3	98.7	99	97.9
No	1.8	1.5	2.7	2.4	7.7	1.3	1.0	2.1
XXX syndrome								
Yes	95.4	96.3	94.7	94.3	92.3	97.5	97.4	95.8
No	4.6	3.7	5.3	5.7	7.7	2.5	2.6	4.2
							(101)

A 4.31. Physician Feels Legally Bound to Reveal Information to Parents, Although Would Prefer to Withhold It (%) (Q15 #1)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	35.1 20.4 44.4	41.8 13.2 45.1	41.7 16.2 42.1	14.7	10.1	52.5 14.3 33.3	52.8 13.9 33.3	45.1 15.1 39.8 (33)

A 4.32. A Physician Should Not Tell Parents About a Fetal Anomaly When of the Opinion It Is Minor (%) (Q15 #20)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	87.4 10.2 2.3	85.5 7.4 7.1	86.9 7.5 5.7	82.6 9.4 8.0	80.9 16.7 2.3	91.2 3.3 5.5	90.5 4.8 4.6	87.5 7.2 5.3 (37)

A 4.33. Early Diagnosis Information on Fetal Sex Should Not Be Disclosed Unless Medically Relevant (%) (Q15 #33)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	42.4 16.9 40.7	60.7 13.7 25.6	49.2 15.5 35.3	33.6 18.4 48.0	33.0 18.4 48.5	36.9 21.2 41.9	51.9 12.2 35.9	47.9 15.6 36.5 (47)

 $p \le 0.01$

A 4.34. There Is a Danger That Results from Predisposition Testing Will Be Used for Discriminatory Purposes (%) (Q15 #3)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	5.8 11.0 83.2	16.3 17.1 66.6	12.7	8.6 14.7 76.7		7.5 15.2 77.4	9.1 13.0 78.0	10.8 13.6 75.6 (25)

A 4.35. With Increasing Refinement in PND Procedures, Conditions Which We Would Otherwise Consider Normal and Accept as Part of Life Are Now Seen as Pathological (%) (Q15 #7)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	17.2 27.5 55.4	26.8 26.2 46.9	22.9 27.9 49.2	33.7	16.1 26.8 57.1	21.4 26.0 52.6	25.1 21.1 53.8	22.9 26.2 50.9 (38)

A 4.36. Use of PND Makes Us More and More Intolerant of the Smallest Anomaly in a Fetus or Child (%) (Q15 #19)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	34.2 22.4 43.4	25.5 16.0 58.5		39.5 20.5 40.0	23.0 16.8 60.1	27.8 17.9 54.3	39.1 18.8 42.1	32.5 18.5 49.0 (35)

 $p \le 0.01$

A 4.37. Giving Birth Intentionally to a Child with a Genetic Defect at a Time When Both PND and Abortion Are Available Is Socially Irresponsible (%) (Q15 #28)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally	82.6 8.8 8.5	54.1 18.5 27.4	14.4		10.3	10.4	75.7 9.5 14.8	71.0 12.7 16.3 (45)

A 4.38. The Success of PND Is Best Measured by Reductions in the Costs of Services for the Care of Children with Genetic Anomalies (%) (Q15 #8)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	47.5 28.0 24.4	38.9 28.7 32.4			61.2 22.4 16.5	53.0 17.8 29.2	51.1 24.5 24.5	47.2 24.7 28.1 (37)

A 4.39. It Would Be Justified to Enact Laws to Control the Spread of Genes Causing Severe Diseases (%) (Q15 #31)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	80.6 10.3 9.1	62.9 15.6 21.5	13.5	78.6 9.7 11.7	13.9	76.8 10.6 12.6	75.9 13.3 10.8	73.6 13.0 13.5 (58)

 $p \le 0.01$

A 4.40. The Handicapped Should Be Consulted During the Development of Policies Concerning PND (%) (Q15 #14)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	17.9 28.8 53.3	21.3	22.8 23.5 53.7	18.5 26.6 54.9	27.8 22.8 49.5	18.6 24.0 57.4	20.0 26.5 53.6	22.4 24.5 53.1 (42)

A 4.41. In General, Women Put Too Much Faith in PND (%) (Q15 #18)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	49.3 35.7 15.0	35.1 33.8 31.1	32.8	51.0 34.4 14.6	32.5 41.4 26.1	51.2 30.5 18.2	57.1 32.7 10.1	46.9 33.4 19.7 (57)

p ≤ 0.01

A 4.42. Funding Priorities* (Q13)

		•						
	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Primary prevention								
Nutritional assistance	2.1	2.6	2.4	2.2	2.4	2.5	2.5	2.4
Multidisciplinary teams Information on harmful effects of smoking and	2.4	2.7	2.9	2.7	2.4	2.5	2.8	2.7
alcohol	2.3	3.0	2.5	2.4	2.5	2.2	2.3	2.5
Screening								
Blood tests	5.3	4.6	4.5	4.6	4.9	4.9	4.4	4.7
Improving cytogenetics Improving obstetrical	5.5	4.6	5.2	5.6	5.5	5.0	5.3	5.2
ultrasound	5.3	4.8	5.2	5.0	4.9	5.2	5.1	5.1
Developing services for the treatment of								
infertility	5.1	5.2	5.2	5.4	5.3	5.4	5.5	5.3 (119)

^{*} Doctors had to rank various items in order of importance from 1 (most important) to 7 (least important).

A 4.43. If It Were Possible to Identify All Cystic Fibrosis Carriers, Systematic Screening of the Entire Population for the Condition Would Be Desirable (%) (Q15 #34)*

	ATL	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	29.6 31.9 38.6	25.3 22.9 51.8	17.9 27.9 54.1	30.9 21.2 47.9	25.2 25.7 49.1	18.6 22.7 58.7	24.0 24.5 51.5 (523)

* This question was not asked in Quebec.

A 4.44. Importance Attributed to Evaluating the Risk of Exposure to Mutagens and Teratogens (%) (Q14)*

	ATL	ONT	MAN	SASK	ALB	BC/NWT	CAN
Not important Moderately important Very important	10.9 32.6 56.6	14.6 29.1 56.3	10.0 26.5 63.6	9.0 30.7 60.3	15.0 27.3 57.7	14.7 18.5 66.8	13.7 26.5 59.8 (611)

 $p \le 0.01$

* This question was not asked in Quebec.

A 4.45. PND Cannot Be Considered a Priority When Only 3% of Children Are Born with an Anomaly While a Much Larger Proportion Born in Good Health Develop Serious Handicaps Caused by Social and Economic Conditions (%) (Q15 #26)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	25.8 21.9 52.3	45.3 18.2 36.5			26.5 14.5 59.0	31.7 16.3 52.0	35.7 18.9 45.4	35.5 18.4 46.1 (28)

A 4.46. In Carrying Out Their Work, Physicians Must Not Consider the Costs of Medical Services (%) (Q15 #11)

	ATL	QUE	ONT	MAN	SASK	ALB	BC/NWT	CAN
Do not agree Moderately agree Totally agree	87.4 10.1 2.5	58.7 21.3 20.1	11.4	77.7 10.2 12.1	13.3	78.0 9.8 12.2	74.2 16.1 9.7	73.9 13.6 12.5 (25)

p ≤ 0.01

Chapter 5. Frequency Tables, by Medical Specialty

A 5.1. Number of Ultrasound Scans Considered Appropriate in the Course of a Normal Pregnancy (%)

n	GPs 1 962	Obste- tricians 373	Paedia- tricians 495	Radiol- ogists 242	CAN 3 072
0 ultrasound	22.8	11.8	18.2	8.2	19.6
1	67.4	62.0	47.3	59.1	63.0
2	9.3	24.3	30.1	30.1	16.0
3	0.5	1.9	4.4	2.6	1.4 (64)

Note: Figures in bold type are statistically significant at $p \le 0.1$.

A 5.2. Acceptability of Various Reasons for Using Ultrasound (%)

	GPs 1 962	Obste- tricians 373	Paedia- tricians 495	Radiol- ogists 242	CAN 3 072
Screening for malformations					
Not justified	25.7	12.1	16.6	10.3	21.4
Moderately justified	20.1	12.9	13.6	10.2	17.4
Totally justified	54.1	75.0	69.8	79.5	61.1 (63)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
The woman decides Exam not important Do not accept refusal and suggest sees another	67.8 30.4	78.7 18.0	70.4 25.1	83.2 11.1	70.7 26.5
doctor	1.8	3.3	4.5	5.9	2.8 (86)

A 5.4. Ultrasound Should Be Subject to a Written Prior Agreement from the Patient (%) (Q15 #13)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree Moderately agree	56.9 18.4	71.3 12.3	42.3 17.5	87.9 6.1	58.7 16.5
Totally agree	24.8	16.3	40.3	6.0	24.7 (31)

A 5.5. Perception of Reliability of Obstetrical Ultrasound to Detect Malformations at 16-20 Weeks' Gestation (%) (Q4)

	GPs 1 962	Obste- tricians 373	Paedia- tricians 495	Radiol- ogists 242	CAN 3 072
Anencephaly					
Not reassured	5.9	2.7	4.1	1.3	4.9
Moderately reassured	8.2	3.0	8.1	0.9	7.0
Totally reassured	85.9	94.2	87.8	97.8	88.1 (54)
Spina bifida					
Not reassured	30.5	14.9	22.2	10.0	25.7
Moderately reassured	36.2	33.2	37.1	30.1	35.5
Totally reassured	33.3	51.9	40.7	59.9	38.8 (58)
Hydrocephaly					
Not reassured	30.1	23.9	25.8	12.2	27.3
Moderately reassured	28.4	27.7	30.2	18.5	27.8
Totally reassured	41.5	48.4	44.0	69.3	44.9
					(60)
Heart malformations					
Not reassured	55.5	47.0	62.4	44.4	54.7
Moderately reassured	30.3	33.1	27.7	39.0	30.9
Totally reassured	14.2	20.0	9.9	16.6	14.4
					(64)
Trisomy 21 (without structural malformations)					
Not reassured	88.5	91.2	87.0	73.9	87.4
Moderately reassured	8.7	6.9	9.6	18.6	9.4
Totally reassured	2.8	1.9	3.4	7.5	3.2
					(63)

A 5.6. Age at Which Amniocentesis Should Be Available to Women Irrespective of Present Policies (%) (Q5A)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
≤34 years	7.3	8.7	17.5	16.0	9.7
35 years	61.5	69.5	62.2	54.6	62.1
36-39 years	17.2	16.3	10.5	13.1	15.7
≥40 years	8.4	3.1	4.8	8.7	7.2
Never	5.5	2.4	5.0	7.6	5.2
					(74)

A 5.7. Age at Which CVS Should Be Available to Women Irrespective of Present Policies (%) (Q5B)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
≤34 years	6.7	6.7	13.7	12.8	8.3
35 years	51.3	57.1	49.1	47.9	51.4
36-39 years	16.9	18.2	11.8	9.4	15.7
≥40 years	11.6	8.4	8.8	15.0	11.1
Never	13.3	9.5	16.6	14.9	13.5 (208)

p ≤ 0.01

A 5.8. Acceptability of Reasons for Using Amniocentesis (%) (Q8)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Anxiety					
Not justified	59.0	53.3	43.4	43.6	54.6
Moderately justified	23.0	20.7	26.5	27.7	23.7
Totally justified	18.0	26.0	30.1	28.7	21.7 (37)
Freedom of choice (age 33)					
Not justified	61.2	51.0	50.9	49.1	57.4
Moderately justified	19.9	21.5	23.1	21.8	21.9
Totally justified	18.9	27.6	25.9	29.1	21.9 (51)
Access without criteria (public health system)					
Not justified (1-2)	78.9	77.6	63.2	68.5	75.4
Moderately justified (3)	9.8	8.7	12.8	12.3	10.4
Totally justified (4-5)	11.3	13.7	24.0	19.2	14.2 (32)
If abortion is refused					
Not justified	52.3	35.4	51.7	60.8	50.8
Moderately justified	13.4	9.4	15.0	11.8	13.0
Totally justified	34.4	55.2	33.3	27.4	36.2 (42)

A 5.9. Physician's Attitude When a Woman Is Hesitant About Amniocentesis (%) (Q6-Q7)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
36-year-old woman					
Recommends procedure Does not recommend	41.3	49.9	55.2	51.4	45.4
procedure Recommends a	19.8	8.1	7.8	5.9	15.4
screening ultrasound	16.2	15.8	22.2	35.4	18.6
Other	27.1	34.9	17.8	9.5	25.1
					(58)
38-year-old woman					
Recommends procedure Does not recommend	60.1	66.0	69.6	63.9	62.6
procedure	9.3	4.3	4.9	4.2	7.6
Recommends a					
screening ultrasound	10.7	7.9	13.0	24.9	11.8
Other	23.2	27.3	14.9	9.0	21.2
					(51)

p ≤ 0.01

A 5.10. Fear of Lawsuits Makes Us Use PND More Often Than Would Be Medically Indicated (%) (Q15 #32)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	27.3	36.2	30.8	19.4	28.3
Moderately agree	13.7	15.1	24.2	17.7	15.9
Totally agree	59.0	48.7	45.0	62.9	55.8 (50)

A 5.11. Acceptability of Predisposition Tests (%) (Q12A)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Diabetes					
In utero	5.5	14.3	7.9	12.2	7.5
Childhood	66.3	66.8	66.5	70.4	66.7
Adulthood	17.6	12.1	13.3	7.3	15.5
Never	10.5	6.9	12.3	10.1	10.3
					(18)
Schizophrenia					
In utero	14.5	26.9	19.1	22.3	17.4
Childhood	44.1	45.4	39.4	46.9	43.7
Adulthood	19.0	14.4	17.8	11.2	17.6
Never	22.4	13.3	23.7	19.6	21.3
					(34)
Alzheimer's disease					
In utero	6.4	13.0	11.2	12.9	8.5
Childhood	17.6	22.6	15.9	22.5	18.3
Adulthood	43.2	42.3	41.9	37.7	42.4
Never	32.8	22.1	31.0	27.0	30.7
					(33)

A 5.12. Acceptability of Reasons for Performing Predisposition Tests (%) (Q12B)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Diabetes					
To prevent births	2.2	4.8	3.3	7.4	3.1
Early treatment	59.6	63.6	55.3	68.1	60.1
Preventive counselling	30.9	26.6	31.2	17.6	29.4
None	7.3	5.0	10.2	7.0	7.5
					(26)
Schizophrenia					
To prevent births	14.4	23.3	17.9	19.8	16.5
Early treatment	46.9	46.5	43.0	49.1	46.4
Preventive counselling	17.7	18.6	18.8	15.0	17.8
None	21.0	11.6	20.3	16.1	19.3
					(37)
Alzheimer's disease					
To prevent births	5.5	10.6	9.1	9.5	7.0
Early treatment	28.9	33.2	30.2	34.4	30.1
Preventive counselling	30.4	31.8	29.3	26.8	30.1
None	35.2	24.4	31.4	29.3	32.8
					(54)

A 5.13. Predetermining an Embryo's Sex by Chromosome Selection (%) (Q15 #9)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Not acceptable	74.4	63.8	71.8	69.0	72.3
Moderately acceptable	11.7	14.1	16.1	13.8	12.9
Totally acceptable	13.9	22.1	12.1	17.1	14.9 (33)

A 5.14. Self-Prescribed Tests to Determine the Sex of the Fetus (%) (Q15 #25)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Not acceptable	67.5	67.1	75.3	71.5	69.0
Moderately acceptable	18.4	16.4	15.3	16.1	17.5
Totally acceptable	14.1	16.5	9.4	12.4	13.5 (23)

A 5.15. Perception of Parents' Difficulties When Offspring Have Various Conditions (%) (Q9A, Q10)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Behaviour problems					
Not difficult	6.1	8.1	12.5	6.7	7.4
Moderately difficult	28.2	30.8	35.8	28.8	30.2
Very difficult	65.0	61.1	51.8	64.5	62.4
					(21)
Cleft lip and palate					
Not difficult	23.6	28.0	28.8	12.6	24.1
Moderately difficult	26.7	30.0	29.7	20.9	27.1
Very difficult	49.7	42.0	41.6	66.5	48.8
-					(16)

A 5.16. I Could Not Accept the Idea of Having a Child with Trisomy 21 (%) (Q15 #30)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	44.6	25.2	38.9	28.1	40.1
Moderately agree	20.8	15.4	18.7	20.1	19.8
Totally agree	34.6	59.4	42.3	51.9	40.2 (57)

A 5.17. Acceptability of Abortion for Certain Conditions (%) (Q11)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Trisomy 21 (without structural					
malformations) Not acceptable	36.6	18.5	29.5	29.2	32.7
Moderately acceptable	17.4	11.2	14.4	18.4	16.2
Totally acceptable	46.0	70.3	56.1	52.4	51.1
Totally acceptable	40.0	70.5	50.1	52.4	(37)
Duchenne muscular dystrophy					, ,
Not acceptable	37.8	19.8	24.8	24.7	32.5
Moderately acceptable	16.6	18.1	13.2	16.4	16.2
Totally acceptable	45.7	62.1	62.1	58.9	51.3
					(27)
Huntington's disease					
Not acceptable	38.8	22.3	26.6	29.4	34.1
Moderately acceptable	14.3	15.0	16.5	13.7	14.7
Totally acceptable	47.0	62.7	56.9	56.9	51.3
					(34)
Severe heart malformations					
Not acceptable	42.7	27.0	32.3	24.5	37.7
Moderately acceptable	19.8	18.9	18.7	13.7	19.1
Totally acceptable	37.4	54.1	49.1	61.8	43.2
,					(58)

Α	5.17.	(cont'd)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Cystic fibrosis					
Not acceptable	48.9	33.7	34.9	38.3	44.0
Moderately acceptable	19.0	21.7	15.3	17.8	18.6
Totally acceptable	32.0	44.6	49.9	43.9	37.4
,					(23)
Spina bifida					
Not acceptable	51.9	35.9	43.6	41.2	47.8
Moderately acceptable	23.4	22.8	21.3	21.8	22.8
Totally acceptable	24.7	41.3	35.1	37.0	29.3
					(27)
Phenylketonuria					
Not acceptable	66.2	53.4	63.2	49.1	62.9
Moderately acceptable	15.7	18.9	12.0	15.7	15.5
Totally acceptable	18.0	27.7	24.8	35.2	21.6
					(43)
Turner's syndrome					
Not acceptable	61.2	51.0	65.3	47.7	59.6
Moderately acceptable	18.9	20.4	19.2	21.9	19.4
Totally acceptable	19.8	28.5	15.5	30.3	21.0
					(46)
Klinefelter's syndrome					
Not acceptable	67.6	48.6	61.9	53.1	63.2
Moderately acceptable	18.2	23.5	19.5	24.6	19.5
Totally acceptable	14.2	28.0	18.5	22.3	17.2
					(27)
XYY syndrome					
Not acceptable	68.3	46.7	64.5	54.1	64.0
Moderately acceptable	18.2	25.2	18.7	25.3	19.7
Totally acceptable	13.4	28.1	16.8	20.6	16.3
					(31)
XXX syndrome					
Not acceptable	68.9	48.8	64.2	54.2	64.6
Moderately acceptable	18.2	23.7	19.3	24.6	19.5
Totally acceptable	12.9	27.4	16.5	21.2	15.9
					(24)

A 5.17. (cont'd)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Lobster claw deformity					
Not acceptable	76.1	59.3	76.2	69.4	73.6
Moderately acceptable	16.0	22.2	12.2	14.2	16.0
Totally acceptable	7.9	18.5	11.6	16.4	10.4 (29)

A 5.18. Elective Abortion Is Less Acceptable Than Abortion of a Fetus with an Anomaly (%) (Q15 #17)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	34.2	31.5	26.2	24.2	31.8
Moderately agree	12.4	11.3	11.8	8.9	11.9
Totally agree	53.4	57.3	62.0	66.9	56.4 (49)

 $p \le 0.01$

A 5.19. Aborting a Fetus with an Anomaly Is More Justifiable in the First Than in the Second Trimester of Pregnancy (%) (Q15 #21)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	38.9	52.0	36.0	35.8	39.7
Moderately agree	15.0	9.0	10.5	14.8	13.5
Totally agree	46.2	39.0	53.5	49.4	46.7 (49)

A 5.20. Aborting a Fetus with a Minor Anomaly Is Justifiable (%) (Q15 #12)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	70.5	31.5	79.3	66.9	70.6
Moderately agree	15.6	17.2	12.0	16.7	15.3
Totally agree	13.9	21.4	8.7	16.4	14.1 (29)

A 5.21. A Physician Must Be Able to Resist Some Abortion Requests When of the Opinion the Anomaly is Minor (%) (Q15 #2)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	25.9	28.3	15.8	20.6	24.1
Moderately agree	14.0	14.5	8.9	12.4	13.1
Totally agree	60.1	57.1	75.3	66.9	62.7 (30)

 $p \le 0.01$

A 5.22. Physicians, Not Parents, Should Decide Which Fetal Anomalies Warrant Pregnancy Termination (%) (Q15 #6)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	60.5	63.5	53.1	58.3	59.5
Moderately agree	25.1	21.1	25.8	27.0	24.9
Totally agree	14.4	15.4	21.1	14.7	15.6 (30)

A 5.23. With Respect to Abortion, Parents Have an Absolute Right to Freedom of Choice (%) (Q15 #4)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	38.2	27.5	33.2	34.1	35.8
Moderately agree	14.2	11.9	18.2	15.1	14.6
Totally agree	47.6	60.6	48.6	50.8	49.6 (17)

A 5.24. Should Parents Be Told if a Fetus Has a Sex Chromosome Anomaly? (%) (Q9B)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
XYY syndrome					
Yes	95.2	95.9	91.9	90.2	94.4
No	4.8	4.1	8.1	9.8	5.6
XXY syndrome					
Yes	98.2	99.0	97.1	95.0	97.9
No	1.8	1.0	2.9	5.0	2.1
XXX syndrome					
Yes	96.1	97.2	95.1	92.0	95.8
No	3.9	2.8	4.9	8.0	4.2

 $p \le 0.01$

A 5.25. Physician Feels Legally Bound to Reveal Information to Parents, Although Would Prefer to Withhold It (%) (Q15 #1)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	48.9	44.8	36.4	31.7	45.1
Moderately agree	14.6	14.9	14.0	21.9	15.1
Totally agree	36.5	40.3	49.7	46.4	39.8 (33)

A 5.26. A Physician Should Not Tell Parents About a Fetal Anomaly When of the Opinion It Is Minor (%) (Q15 #20)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	89.6	87.5	83.5	79.0	87.5
Moderately agree	6.7	5.6	9.1	10.4	7.2
Totally agree	3.8	6.9	7.4	10.6	5.3 (37)

A 5.27. Early Diagnosis Information on Fetal Sex Should Not Be Disclosed Unless Medically Relevant (%) (Q15 #33)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	47.1	62.6	44.7	38.3	47.9
Moderately agree	15.6	10.4	18.5	17.2	15.6
Totally agree	37.2	26.9	36.7	44.4	36.5 (47)

 $p \le 0.01$

A 5.28. With Increasing Refinement in PND, Conditions That We Would Otherwise Consider Normal and Accept as Part of Life Are Now Seen as Pathological (%) (Q15 #7)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	19.7	26.7	28.7	30.9	22.9
Moderately agree	24.6	28.1	30.0	28.2	26.2
Totally agree	55.7	45.2	41.3	40.8	50.9 (38)

A 5.29. Giving Birth Intentionally to a Child with a Genetic Defect at a Time When Both PND and Abortion Are Available Is Socially Irresponsible (%) (Q15 #28)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	76.6	62.9	63.1	53.6	71.0
Moderately agree	11.1	14.1	16.5	16.0	12.7
Totally agree	12.3	23.0	20.4	30.4	16.3
					(45)

A 5.30. It Would Be Justified to Enact Laws to Control the Spread of Genes Causing Severe Diseases (%) (Q15 #31)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	76.2	64.6	76.3	60.2	73.6
Moderately agree	12.4	14.6	11.0	18.9	13.0
Totally agree	11.4	20.8	12.6	20.9	13.5 (58)

 $p \le 0.01$

A 5.31. Importance of Assessing Exposure to Mutagenic and Teratogenic Hazards (%) (Q14)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	13.2 24.9	17.6	11.6	16.9 29.0	13.7 26.5
Moderately agree Totally agree	61.9	36.7 45.6	25.7 62.7	54.1	59.8 (28)

A 5.32. PND Cannot Be Considered a Priority When Only 3% of Children Are Born with an Anomaly While a Much Larger Proportion Born in Good Health Develop Serious Handicaps Caused by Social and Economic Conditions (%) (Q15 #26)

	GPs	Obste- tricians	Paedia- tricians	Radiol- ogists	CAN
Do not agree	30.0	44.1	45.7	46.1	35.5
Moderately agree	18.1	21.8	17.3	18.3	18.4
Totally agree	51.9	34.0	37.0	35.7	46.1 (28)

Chapter 6. (a) Frequency by Religion

	Catholic 860	Anglican 304	Church 401	Protes- tant 472	Jewish 219	Oriental 74	None 556	Total 3 072
Screening for malformations								
Not justified	15.8	17.1	22.9	34.7	14.4	24.6	21.9	21.4
Moderately justified	15.1	24.0	14.7	18.7	13.8	27.5	17.7	17.5
Totally justified	0.69	58.9	62.4	46.6	71.8	47.8	60.4	61.2

	Catholic	Catholic Anglican	Church	tant	Jewish	Oriental None	None	Total
The woman decides	67.7	71.6	70.8	62.4	58.3	57.6	73.8	68.6
Exam not important	24.6	20.3	23.1	31.9	38	37.9	20	24.8
Do not accept refusal and suggest to								
see another doctor	5.5	2.3	0.4	2.0	2.2	3.0	1.5	2.7
								(145)*

	Catholic	Anglican	United Church	Protes- tant	Jewish	Jewish Oriental	None	Total
34 vears	8.1	10.4	11.1	9.6	12.2	11.4	9.6	9.7
Vears	60.3	64.2	68.7	26.7	70.2	57.8	64.0	62.1
36 years and +	24.5	22.1	17.5	23.7	23.0	15.5	30.0	22.3
Never	7.1	6.0	2.6	9.7	3.2	2.1	0.7	5.2
								(133)

	Catholic	Anglican	United	Protes- tant	Jewish	Oriental	None	Total
34 years	6.3	7.1	9.7	10.3	10.8	5.7	8.7	8.3
35 years	44.0	62.1	61.8	46.7	62.3	52.2	52.1	51.5
36 years and +	29.3	23.3	21.4	25.0	30.7	21.6	35.2	. 26.6
Never	20.4	7.5	7.2	18.1	18.4	5.3	6.9	13.0
								(592)

	Catholic	Catholic Anglican	United	Protes- tant	Jewish	Oriental	None	Total
f abortion is refused								
Not justified	52.0	50.6	45.7	53.5	41.0	55.6	55.2	50.5
Moderately justified	13.1	12.4	14.2	15.2	12.5	14.5	6	13.1
Totally justified	34.9	36.9	40.2	31.3	46.6	29.9	35.5	36.1
								(102)

	Catholic	Anglican	United	Protes- tant	Jewish	Oriental	None	Total
36-vear-old woman								
Recommends procedure	50.8	47.4	42.2	40.0	53.6	44.7	42.7	45.4
Does not recommend procedure	16.4	15.0	13.6	19.5	7.7	1.9	15.2	15.4
Becommends a screening ultrasound	19.4	16.5	18.8	20.0	17.9	31.5	16.5	18.5
Other	16.7	27.8	30.6	23.7	26.6	26.3	30.8	25.1
								(116)
38-vear-old woman								
Recommends procedure	65.5	62.7	66.7	56.6	71.6	68.7	9.69	62.7
Does not recommend procedure	10.4	6.2	4.4	8.8	2.0	0.0	7.3	7.6
Recommends a screening ultrasound	12.3	11.6	7.2	16.6	12.0	20.5	8.9	11.7
Other	15.2	22.9	25.3	20.8	19.1	12.9	27.2	21.2
								(109)

	Catholic	Anglican	Church	tant	Jewish	Oriental	None	Total
4								
Do not agree	32.6	24.7	31.9	19.7	29.7	35.5	29.9	28.4
Moderately agree	16.0	17.2	12.0	18.9	16.3	9.7	16.6	15.8
Totally agree	51.3	58.1	56.1	61.4	54.0	54.8	73.7	55.7
) :	2		(107)

en the Female Partner Has a	
urrogate Motherhood to Couples Wh	
A 6.8. It is Acceptable to Recommend S	Dominant Genetic Disorder (%) (Q15 #16)

	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Do not agree Moderately agree Totally agree	38.4 27.2 34.4	30.8 29.8 39.3	27.9 29.4 42.7	29.8 36.4 33.8	34.5 47.7	31.1 33.1 35.8	23.2 22.3 54.5	30.6 28.9 40.5
p ≤ 0.01								

A 6.9. It is Acceptable to Recommend Artificial Insemination to Couples When the Male Partner Has a Dominant Genetic Disorder (%) (Q15 #22)

	Catholic	Anglican	United	Protes- tant	Jewish	Oriental	None	Total
Do not agree	14.1	9.5	3.5	10.4	2.8 17.4	15.0 28.0	5.0	9.3
Totally agree	65.4	79.6	85.5	71.9	79.8	57.0	82.5	74.5 (92)
p ≤ 0.01								

	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Aggressiveness								
Not difficult	13.2	9.7	10.9	12.6	15.3	24.2	6 1	116
Moderately difficult	35.0	26.9	29.8	34.6	24.5	37.6	35.5	32.6
Very difficult	51.9	63.4	59.3	52.8	60.1	38.2	58.4	55.6
								(06)
Learning disabilities								
Not difficult	15.8	17.6	17.9	21.1	8.7	10.8	14.3	16.2
Moderately difficult	35.0	41.5	33.4	39.1	42.3	515	40.5	8
Very difficult	49.3	40.9	48.8	39.9	49.0	37.7	45.2	45.4
								(63)
Female sterility								
Not difficult	52.6	55.9	64.1	63.6	47.7	40.7	70 9	70
Moderately difficult	30.3	27.5	24.1	22.9	27.6	41.4	18.6	26.0
Very difficult	17.1	16.7	11.8	13.6	24.8	17.8	10.5	15.0
								(73)
Male sterility								
Not difficult	54.7	56.9	65.1	9.29	49.5	43.0	73.5	61.2
Moderately difficult	30.0	26.7	23.4	22.3	27.9	42.5	18.3	25.7
Very difficult	15.2	16.4	11.5	10.1	22.6	14.6	8.2	13.2
								(70)

	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Hypogonadism	426	42.6	53.5	49.8	32.8	35.6	53.9	46.4
Moderately difficult	34.8	36.8	28.6	29.1	32.7	44.4	27.3	32.1
Moderately difficult	22.6	20.7	17.9	21.1	34.5	20.0	18.8	21.5 (88)
Severe cleft lin and palate								
Not difficult	20.3	25.0	24.7	30.3	18.3	22.1	25.1	24.0
Moderately difficult	27.0	31.4	25.5	26.3	15.9	29.5	29.1	27.1
Very difficult	52.7	43.6	49.7	43.4	65.8	48.4	45.8	48.9 (77)
Lobster claw deformity						1		
Not difficult	23.4	27.3	36.3	39.0	18.8	25.2	29.4	29.1
Moderately difficult	29.8	34.3	30.6	24.7	25.3	21.8	31.7	29.8
Very difficult	46.7	38.4	33.1	36.3	55.9	53.0	38.9	41.1
Intellectual impairment					1		,	,
Not difficult	11.6	16.4	13.0	17.6	7.8	15.3	œ. E	13.4
Moderately difficult	21.2	27.9	28.8	26.8	17.5	30.0	26.8	25.6
Very difficult	67.1	55.7	58.3	55.6	74.7	54.6	59.4	61.0

	Catholic	Anglican	United Church	Protes- tant		Jewish Oriental None	None	Total
araplegia								
Not difficult	4.2	8.7	5.5	6 6	<u>.</u>	0 1	27	ù
Alternation of the state of the		. !)	į		3		,
oderately difficult	. / 8	18.9	9.11	10.9	8.2	9.5	11.3	+
/ery difficult	87.1	72.4	82.9	80.0	90.5	88.6	85.0	83.6
								(81)

	Catholic	Catholic Anglican	United Church	Protes- tant	Jewish	Oriental None	None	Total
Do not agree	41.3	42.0	41.7	47.8	20.6	40.8		40
Moderately agree	15.8	25.5	21.3	21.4	21.0	14.3	20.3	19
lotally agree	42.9	32.5	36.9	30.7	58.4	44.9	46.6	40.1
								(116)

	Catholic	Anglican	United	Protes- tant	Jewish	Oriental	None	Total
Trisomy 21 (without structural								
malformations)								
Not acceptable	40.9	34.9	25.5	44.4	13.6	35.1	17.8	32.7
Moderately acceptable	13.7	21.7	18.9	13.6	20.0	17.2	16.9	16.2
Totally acceptable	45.4	43.4	55.6	42.0	66.5	47.7	65.3	51.0 (98)
Duchenne muscular dystrophy							!	
Not acceptable	44.2	27.4	23.6	44.4	9.4	28.7	19.7	32.6
Moderately accentable	17.4	17.0	18.4	12.8	14.0	29.5	13.6	15.9
Totally acceptable	38.4	55.5	58.0	42.8	9.92	41.8	2.99	51.5 (87)
Huntington's disease								
Not acceptable	44.9	27.7	25.7	46.2	8.0	30.6	22.4	34.1
Moderately acceptable	16.5	11.6	14.8	10.7	14.7	28.3	14.1	14.6
Totally acceptable	38.6	8.09	59.5	43.0	75.5	41.2	63.5	51.3 (93)
Severe heart malformations								
Not acceptable	45.3	27.1	33.4	46.7	19.9	29.4	31.7	37.7
Moderately acceptable	17.1	26.8	22.3	17.3	21.5	18.7	18.2	19.2
Totally acceptable	37.6	46.1	44.3	36.0	58.6	52.0	50.1	43.1
								(118)

6.12. (cont'd)

	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Cystic fibrosis Not acceptable	54.8	42.4	37.0	54.7	23.9	47.0	0 80	0 88
Moderately acceptable	18.0	16.4	22.4	13.1	18.0	25.8	22.5	18.6
Totally acceptable	27.2	41.2	40.5	32.1	58.1	27.2	48.5	37.4 (83)
Spina bifida	1							
ivot acceptable Moderately acceptable	58.2	42.5 28 1	48.4	50.7	29.6	57.9	34.6	48.0
Totally acceptable	23.6	29.5	27.4	26.6	45.2	22.4	36.3	23.0
								(87)
Phenylketonuria								
Not acceptable	67.1	60.1	59.2	73.1	50.7	58.2	55.1	63.1
Moderately acceptable	14.4	18.4	16.1	11.8	16.7	13.2	18.6	15.3
lotally acceptable	18.5	21.5	24.7	15.1	32.6	28.5	26.3	21.6
								(103)
Turner's syndrome								
or acceptable	9.99	56.5	49.8	68.8	44.3	62.4	54.9	59.8
Moderately acceptable	16.8	21.4	24.7	13.7	26.2	15.0	20.1	19.2
lotally acceptable	16.6	22.1	25.5	17.4	29.5	22.6	25.0	21.0
								(107)

	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Klinefelter's syndrome	69.5	54.7	58.6	70.2	46.5	62.6	60.5	63.5
Moderately acceptable	18.5	24.2	24.6	14.8	22.0	14.2	18.8	19.4
Totally acceptable	12.0	21.1	16.9	14.9	31.5	23.2	20.7	17.1 (88)
XVV syndrome								
Not acceptable	70.8	54.3	57.7	70.7	47.8	9.07	6.19	64.2
Moderately acceptable	17.3	28.8	26.0	14.1	24.6	10.7	18.0	19.6
Totally acceptable	11.9	17.0	16.3	15.2	27.7	18.7	20.1	16.2 (92)
VVV evndrome								
Not acceptable	2.69	59.0	59.3	8.69	49.2	9.69	62.8	64.8
Moderately acceptable	18.4	22.8	24.5	16.3	22.4	16.2	18.2	19.5
Totally acceptable	11.9	18.2	16.1	13.9	28.4	14.2	19.0	15.8 (84)
Lobster claw deformity			1			1	7	1
Not acceptable	76.8	68.2	76.5	17.4	62.4	70.3	5.17	2.0
Moderately acceptable	15.4	19.1	16.5	12.9	19.0	12.9	15.7	15.9
Totally acceptable	7.8	12.7	7.1.	9.7	18.7	16.8	13.0	10.3 (88)

A 6.13. Elective Abortion Is Less Acceptable Than Abortion of a Fetus with an Anomaly (%) (Q15 #17)

	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Do not agree	27.1	26.9	36.0	27.0	47.0	19.1	32.2	31.7
Moderately agree	12.3	14.2	13.4	11.3	13.2	9.7	8.7	11.7
Totally agree	9.09	58.8	50.5	61.7	39.8	71.2	53.0	56.5
								(118)
p ≤ 0.01								

A 6.14. One Must Condemn PND Done with the Deliberate Intention of Terminating the Pregnancy if Results Show an Anomaly (%) (Q15 #10)

			United	Protes-				
	Catholic	Anglican	Church	tant	Jewish	Oriental	None	Total
Do not agree	63.4	75.0	77 1	0 63	07.6	000		1
	1.00	7.07	1./	02.0	0.70	00.0	80.5	72.1
Moderately agree	12.2	0.9	6.6	12.4	6.3	8.0	6.7	6.6
Totally agree	24.5	18.8	12.9	23.8	6.2	23.2	6.7	18.1
								(66)
p < 0.01								
-								

A 6.15. A Physician Must Be Able to Resist Some Abortion Requests When of the Opinion the Anomaly Is Minor (%) (Q15 #2)

	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Do not agree	19.9	27.4	26.5	16.8	38.9	15.2	30.1	24.0
Moderately agree	14.2	16.1	12.0	12.4	13.4	7.6	12.6	13.2
Totally agree	62.9	56.5	61.5	70.8	47.7	77.2	57.3	62.8
								(68)
p ≤ 0.01								

	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental None	None	Total
Do not agree	45.2	34.5	22.9	52.2	16.3	24.3	24.6	35.9
Moderately agree	14.4	14.6	21.0	89.89	10.9	36.0	15.7	14.6
Totally agree	40.4	50.9	56.1	38.9	72.8	39.7	265	49.5
								(78)

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	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Do not agree	51.2	48.4	46.1	39.5	58.8	31.0	51.0	48.4
Moderately agree	13.4	19.6	15.5	17.3	14.9	16.3	13.3	15.6
Totally agree	35.4	32.1	38.4	43.3	26.3	52.7	34.8	36.2
								(107)
$p \leq 0.01$								

and	Total	22.9 26.1 51.0
Normal	None	24.5 31.3 44.2
Consider	Jewish Oriental None	15.4 24.0 60.6
Otherwise	Jewish	33.8 33.1 33.1
Would (Protes- tant	22.5 21.8 55.7
s Which We (a) (Q15 #7)	United	25.5 28.8 45.8
Conditions ological (%	Catholic Anglican	17.9 25.5 56.6
ment in PND, Seen as Path	Catholic	20.9 23.6 55.5
A 6.18. With Increasing Refinement in PND, Conditions Which We Would Otherwise Consider Normal and Accept as Part of Life Are Now Seen as Pathological (%) (Q15 #7)		Do not agree Moderately agree Totally agree

A 6.19. Use of PND Makes Us More and More Intolerant of the Smallest Anomaly in a Fetus or Child (%) (Q15 #19)

			Chited	Protes-				
	Catholic	Anglican	Church	tant	Jewish	Oriental	None	Total
Do not agree	24.1	29.4	38.7	34.9	47.1	28.2	37.2	32.5
Moderately agree	15.3	22.7	18.9	12.5	19.0	8.7	26.5	18.3
Totally agree	9.09	47.9	42.5	52.6	33.9	63.0	36.3	49.2
								(96)
p < 0.01								

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	Catholic	Anglican	United Church	Protes- tant	Jewish	Oriental	None	Total
Do not agree	14.7	14.6	9.5	17.1	12.7	6.1	14.2	13.7
Moderately agree	33.2	28.5	23.0	21.8	19.1	19.0	30.7	26.5
49.5	52.1	56.9	67.5	61.1	68.2	75.0	55.2	59.8
_								(663)

A 6.21. If It Were Possible to Identify All Cystic Fibrosis Carriers, Systematic Screening of the Entire Population for the Condition Would Be Desirable (%) (Q15 #34)

	Catholic	Anglican	United	Protes- tant	Jewish	Oriental	None	Total
Do not agree	26.7	24.6	21.3	29.1	1.3	19.8	22.0	24.0
Moderately agree	21.5	29.0	19.2	25.9	17.7	33.9	26.1	24.4
Totally agree	51.8	46.4	59.5	44.9	64.0	46.3	51.9	51.7
								(577)
p ≤ 0.01								

A 6.22. PND Cannot Be Considered a Priority When Only 3% of Children Are Born with an Anomaly While a Much Larger Proportion Born in Good Health Develop Serious Handicaps Caused by Social and Economic Conditions (%) (Q15 #26)

Catholic Anglican		United	Protes- tant	Jewish	Oriental	None	Total
33.9		0:	29.1	52.8	35.7	41.1	35.3
20.6	16.8 16.1		19.1	16.6	13.1	19.4	18.5
45.4		6.	51.8	30.5	51.2	39.5	46.2
							(88)

(b) Frequency by Degree of Religious Practice

A 6.23. Acceptability of Reasons for Using Ultrasound Scanning (%) (Q3)

	Practising n = 1 262	Occasional n = 865	Non-practising n = 754	Total n = 3 072
Screening for malformations				
Not justified	26.2	17.2	17.7	21.3
Moderately justified	18.5	15.3	18.2	17.5
Totally justified	55.3	67.5	64.1	61.2 (244)

 $p \le 0.01$

A 6.24. Age at Which Amniocentesis Should Be Available to Women Irrespective of Present Policies (%) (Q5A)

	Practising	Occasional	Non-practising	Total
≤34 years	7.8	12.3	9.8	9.7
35 years	56.2	65.0	69.2	62.2
36-39 years	17.4	14.0	14.7	15.7
≥40 years	10.6	4.8	4.8	7.4
Never	8.0	3.9	1.5	5.1
				(259)

A 6.25. Physician's Attitude When a Woman Is Hesitant About Amniocentesis (%) (Q6-Q7)

	Practising	Occasional	Non-practising	Total
36-year-old woman				
Recommends procedure Does not recommend	39.4	51.3	44.3	44.3
procedure Recommends a screening	18.6	12.4	12.7	15.2
ultrasound	19.6	17.5	14.9	17.7
Other	22.5	18.9	28.1	22.9 (244)
38-year-old woman				
Recommends procedure Does not recommend	55.9	69.1	63.5	61.9
procedure Recommends a screening	10.5	4.4	5.8	7.5
ultrasound	13.8	10.5	7.1	11.1
Other	19.7	16.0	23.6	19.6 (236)

p ≤ 0.01

A 6.26. Fear of Lawsuits Makes Us Use PND More Often Than Would Be Medically Indicated (%) (Q15 #32)

	Practising	Occasional	Non-practising	Total
Do not agree	23.8	31.2	34.6	28.9
Moderately agree	14.3	18.6	14.3	15.6
Totally agree	61.9	50.2	51.1	55.5 (236)

A 6.27. I Could Not Accept the Idea of Having a Child with Trisomy 21 (%) (Q15 #30)

	Practising	Occasional	Non-practising	Total
Do not agree	53.6	32.3	28.3	40.6
Moderately agree	18.5	21.2	20.4	19.8
Totally agree	27.9	46.5	51.3	39.6 (245)

A 6.28.	Acceptability	of	Abortion	for	Certain	Conditions	(%)	(Q11))
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	Practising	Occasional	Non-practising	Total
Trisomy 21 (without				
structural malformations)				
Not acceptable	51.5	22.4	13.8	33.0
Moderately acceptable	13.6	18.9	17.6	16.2
Totally acceptable	34.9	58.7	68.5	50.8
				(222)
Duchenne muscular				
dystrophy				
Not acceptable	48.3	24.2	17.4	33.0
Moderately acceptable	14.2	18.6	16.6	16.1
Totally acceptable	37.6	57.2	65.9	50.9
•				(215)
Huntington's disease				
Not acceptable	47.8	27.7	19.4	34.4
Moderately acceptable	12.9	16.4	16.6	14.9
Totally acceptable	39.3	55.9	64.0	50.7
				(222)
Severe heart				
malformations				
Not acceptable	52.8	27.3	23.4	37.5
Moderately acceptable	16.0	22.0	21.8	19.3
Totally acceptable	31.3	50.7	54.9	43.2
				(245)
Cystic fibrosis				
Not acceptable	59.4	35.7	29.4	44.4
Moderately acceptable	14.3	22.6	21.8	18.7
Totally acceptable	26.3	41.8	48.9	36.8
				(209)

A 6.28. (cont'd)

	Practising	Occasional	Non-practising	Total
Spina bifida				
Not acceptable	60.1	42.2	32.8	47.7
Moderately acceptable	19.5	25.6	25.4	22.9
Totally acceptable	20.3	32.3	41.8	29.5
,				(215)
Phenylketonuria				
Not acceptable	73.0	58.3	51.8	63.1
Moderately acceptable	11.8	15.7	21.4	15.5
Totally acceptable	15.2	26.0	26.8	21.4
				(230)
Turner's syndrome				
Not acceptable	72.0	51.3	48.1	59.6
Moderately acceptable	13.6	23.5	25.2	19.6
Totally acceptable	14.4	25.2	26.7	20.8
				(234)
Klinefelter's syndrome				
Not acceptable	73.5	54.0	56.8	63.3
Moderately acceptable	14.2	25.0	23.4	19.8
Totally acceptable	12.3	21.0	19.8	16.9
				(209)
XYY syndrome				
Not acceptable	73.5	55.0	58.2	64.0
Moderately acceptable	15.3	24.8	22.4	20.0
Totally acceptable	11.2	20.2	19.4	16.0
				(214)
XXX syndrome				
Not acceptable	73.1	57.3	58.5	64.6
Moderately acceptable	15.1	23.5	23.9	19.9
Totally acceptable	11.7	19.2	17.6	15.5
				(211)
Lobster claw deformity				
Not acceptable	79.1	70.2	67.2	73.6
Moderately acceptable	12.9	18.7	19.9	16.0
Totally acceptable	8.0	11.1	12.9	10.4
•				(216)

A 6.29. Acceptability of Reasons for Using Predisposition Tests (%) (Q12B)

	Practising	Occasional	Non-practising	Total
Schizophrenia				
Preventing births	11.4	20.4	19.9	16.4
Early treatment	49.1	45.6	43.3	46.5
Preventive counselling	21.2	15.2	15.5	17.9
None	18.3	18.8	21.2	19.2
				(226)

p ≤ 0.01

A 6.30. Aborting a Fetus with a Minor Anomaly Is Justifiable (%) (Q15 #12)

	Practising	Occasional	Non-practising	Total
Do not agree	77.9	68.4	60.5	70.5
Moderately agree	11.3	17.7	19.7	15.4
Totally agree	10.8	13.9	19.8	14.1
				(218)

 $p \le 0.01$

A 6.31. A Physician Must Discuss the Question of Abortion with Alcoholic Women (%) (Q15 #24)

	Practising	Occasional	Non-practising	Total
Do not agree	51.2	34.7	34.1	41.8
Moderately agree	25.0	31.7	26.1	27.3
Totally agree	23.8	33.6	39.8	30.9 (226)

A 6.32. One Must Condemn PND Done with the Deliberate Intention of Terminating the Pregnancy if Results Show an Anomaly (%) (Q15 #10)

	Practising	Occasional	Non-practising	Total
Do not agree	60.1	76.9	84.4	71.6
Moderately agree	12.5	10.0	5.2	9.9
Totally agree	27.4	13.0	10.3	18.6 (227)

A 6.33. A Physician Must Be Able to Resist Some Abortion Requests When of the Opinion the Anomaly is Minor (%) (Q15 #2)

	Practising	Occasional	Non-practising	Total
Do not agree	18.3	25.7	32.2	24.2
Moderately agree	11.7	14.2	15.2	13.4
Totally agree	70.0	60.1	52.6	62.5 (215)

 $p \le 0.01$

A 6.34. Parents Have an Absolute Right to Freedom of Choice with Respect to Abortion (%) (Q15 #4)

	Practising	Occasional	Non-practising	Total
Do not agree	49.1	28.0	24.4	36.3
Moderately agree	11.0	18.3	15.7	14.4
Totally agree	39.9	53.7	59.9	49.3 (206)

A 6.35. With Increasing Refinement in PND, Conditions Which We Would Otherwise Consider Normal and Accept as Part of Life Are Now Seen as Pathological (%) (Q15 #7)

	Practising	Occasional	Non-practising	Total
Do not agree	16.6	26.1	28.1	22.5
Moderately agree	24.1	25.5	30.2	26.1
Totally agree	59.3	48.4	41.7	51.4 (222)

A 6.36. Use of PND Makes Us More and More Intolerant of the Smallest Anomaly in a Fetus or Child (%) (Q15 #19)

	Practising	Occasional	Non-practising	Total
Do not agree	27.3	34.0	39.3	32.5
Moderately agree	14.6	18.0	21.7	17.5
Totally agree	58.1	47.9	38.9	50.0 (224)

 $p \le 0.01$

A 6.37. Giving Birth Intentionally to a Child with a Genetic Defect at a Time When Both PND and Abortion Are Available Is Socially Irresponsible (%) (Q15 #28)

	Practising	Occasional	Non-practising	Total
Do not agree	78.4	64.1	66.1	70.9
Moderately agree	9.9	17.3	12.7	12.9
Totally agree	11.7	18.7	21.3	16.3 (234)

A 6.38. PND Cannot Be Considered a Priority When Only 3% of Children Are Born with an Anomaly While a Much Larger Proportion Born in Good Health Develop Serious Handicaps Caused by Social and Economic Conditions (%) (Q15 #26)

	Practising	Occasional	Non-practising	Total
Do not agree	28.9	37.9	41.0	34.8
Moderately agree	17.9	17.4	21.7	18.8
Totally agree	53.2	44.6	37.2	46.5
				(217)

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Natalie Kishchuk joined the present project team as a researcher, contributing, among other things, her knowledge of English Canada and her expertise in cognitive psychology.

Louise Bouchard has acted as Research Supervisor from the beginning of the Quebec/France project. She has been the linchpin for the project as a whole. Jocelyn Bisson joined the team to direct the field work for this study, bringing his expertise in data analysis. Jean-François Labadie handled data analysis, and Jocelyne Boivin-Ostiguy has acted as a parttime research assistant throughout both projects.

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Notes

- 1. Consensus is determined according to the following rule: it is strong if 75% of physicians or more agree with the statement, moderate if 65% to 75% agree, and weak if 55% to 64% agree. In other instances, we consider that the matter is subject to debate.
- 2. U.S. National Research Council, Committee for the Study of Inborn Errors of Metabolism (1975), Powledge and Fletcher (1979), U.S. National Institute of Child Health and Human Development (1979), Hamerton (1980), Science Council of Canada (1980), Réseau provincial de médecine génétique (1980), U.S. President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research (1983), France, Comité consultatif national d'éthique pour les sciences de la vie et de la santé (1985), Royal College of Physicians of London (1989), Council of Europe (CHBI) (1990).
- 3. This estimated \$3 billion project is the most important scientific project funded by the U.S. federal government since the Apollo Project (March of Dimes 1989).
- 4. For example, the screening program for sickle-cell anaemia among black Americans, introduced in the 1970s, had considerable discriminatory effects. The program was introduced so quickly, with no debate or evaluation and no distinction between carriers and affected individuals, that people's employment and marital rights were ultimately violated. It could therefore be said that more harm than good was done in this case.
- 5. Consider, for instance, the ground covered since the U.S. Surgeon General declared tobacco a major risk factor in 1964. Smoking went rapidly from being acceptable to being deviant behaviour, then a disease.
- 6. To put the issue of malformations in perspective, the incidence of low birthweight (6%) and premature births (6.4%) should be borne in mind. In Quebec

at least, the incidence of low birthweight in the poor neighbourhoods of large cities is 10%-11%. According to some authors, between 10% and 20% of children who manage to survive despite very low birthweight suffer permanent disability.

- 7. The position of the Hastings Center is revealing. "Although we strongly oppose any movement aimed at making diagnosis of sex and selective abortion a part of ordinary medical practice and family planning, we recommend that no legal restrictions be placed on ascertainment of fetal sex. We think such restrictions would be ineffective and impossible to administer, would lead to subterfuge and, more important, would violate our objective of noninterference with parental choice, even when we disagree with that choice. Though we support the right of individual physicians to refuse to perform prenatal diagnosis for sex choice, we also recognize that in special situations, sex choice can appear to parents to be justifiable. We think most couples should not seek such information, however. Discouragement of this use of prenatal diagnosis, by pointing out that the risks and stresses of second-trimester abortions are not trivial, will mean that such cases will at least not be very great in number ..." (Powledge and Fletcher 1979, 172).
- 8. Q16 "Medical school attended," Q18 "Type of practice," Q19 "Province of practice," Q21 "Distance from a genetics centre," Q34 "Ethnic group," and Q35 "Religion."
- 9. Q9 (B), Q17, Q24, and Q25.
- 10. The list of Quebec doctors was prepared from the membership lists of the Association of Obstetricians and Gynaecologists of Quebec, the Association of Paediatricians of the Province of Quebec, the Fédération des omnipraticiens du Québec, and the Association des radiologies. We selected the names of radiologists who billed the Quebec health insurance plan for 100 or more obstetrical ultrasounds in 1988.
- 11. This definition of language is used in other polls and surveys of physicians.
- 12. We excluded 8 Quebec physicians from the sample because they failed to answer at least 50% of the questionnaire; 43 doctors from English Canada were excluded for the same reason.
- 13. See Dillman (1978).
- 14. In Quebec, the questionnaire was accompanied by a covering letter from the Corporation professionelle des médecins du Québec and the relevant medical federations (FMOQ and FMSQ).
- 15. This discrepancy may be explained by the different ways in which various specialties are registered at the CPMQ and at the CMA.
- 16. Statistical analysis showed no significant differences between the provinces that were combined.
- 17. To make the text easier to read, the detailed frequency tables are to be found at Appendix 3. The first digit of the table number indicates the chapter where it is presented, and the second the order in which it is discussed in the text.
- 18. Relatively few French doctors have recently immigrated to Quebec. On the other hand, based on the place where they trained, about 10% of physicians whom we coded as "British" appeared to be recent immigrants from Great Britain. This would mean that the other 90% were English Canadian.

- 19. These questions were not asked in the Quebec/France survey; the results therefore do not include Quebec doctors.
- 20. Ibid.
- 21. Consensus is determined according to the following rule: it is strong if 75% of physicians or more agree with the statement, moderate if 65% to 75% agree, and weak if 55% to 64% agree. In other instances, we consider that the matter is subject to debate.
- 22. The question asked by Wertz and Fletcher (1989b) specified that contrary to Canada, where the Canadian College of Medical Geneticists has issued a set of guidelines no rules limited access to the test ("assume that your clinic has no regulations that would prevent your doing prenatal diagnosis for her"). Our question merely emphasized the unusual nature of the question. In our questionnaire, the woman was 33 years old; in Wertz and Fletcher's questionnaire, the woman was 25.
- 23. Our question and that of Wertz and Fletcher were slightly different. Ours read as follows:
 - 4. A couple had not intended to have another child. They already have three children of the same sex. The woman initially planned to terminate this pregnancy and have a tubal ligation. Her age does not qualify her for prenatal diagnosis. They now request chorionic villus sampling to learn the sex of the child. She will continue the pregnancy only if the results of the text show the fetus to be of the sex opposite to that of her other children.

Wertz and Fletcher's question read as follows:

A couple requests prenatal diagnosis for purposes of selecting the sex of the child. They already have four girls and are desperate for a boy. They say that if the fetus is a girl, they will abort it and will keep trying until they conceive a boy. They also tell you that if you refuse to do prenatal diagnosis for sex selection, they will abort the fetus rather than run the risk of having another girl. (1989b, 15)

- 24. Physicians were asked to answer the open question: "What is your religion?" For analysis purpose, religions were arranged in groups (see Appendix 2) after consultation with the Canadian Centre for Ecumenism.
- $25.\ Physicians$ were asked to indicate whether they practised their religion regularly, occasionally, or never.
- 26. It should be noted that the statistical tests do not "prove" this causal relationship. They only make it possible to assert that it makes sense.
- 27. The problems are the following: aggressiveness, intellectual deficiency, hypogonadism, behavioural problems, male sterility, female sterility, learning disabilities, paraplegia, severe bilateral cleft lip and palate, and lobster claw deformity.
- 28. The anomalies are the following: severe heart defect, lobster claw deformity of the hand, spina bifida, trisomy 21, XYY syndrome, Klinefelter's syndrome (XXY), XXX syndrome, Turner's syndrome (XO), cystic fibrosis, Duchenne muscular dystrophy, Huntington's disease, and phenylketonuria.

- 29. All the variance analyses performed are based on the regression analysis model, in which the impact of each predictor is evaluated by controlling for the effect of other predictors. "F" tests thus make it possible to evaluate the significance of the unique contribution of each variable.
- 30. A Type I error occurs when a difference is considered significant but, in reality, is not.
- 31. In the context of these analyses, the proportion of the variance accounted for by each factor is the specific contribution made by each factor in reducing the unaccounted-for variance.
- 32. That is, when all other sociocultural factors are controlled for, so that all provinces can be compared in terms of those characteristics.
- 33. It should be noted that GPs represented 65.8% of the sample. Small deviations from the mean thus represented substantial disparities.
- 34. This factor, however, presented some problems for analysis. The difficulty stemmed from the correlation that exists in certain provinces, including Quebec, between religion and ethnic origin. For example, the majority of Catholic physicians in Quebec are of French origin. Although this correlation can be partly offset by statistical control, the difficulty increases as the proportion approaches 100%. For instance, if 95% of Catholic physicians are French, no matter how much one controls for religion, the fact remains that French physicians are Catholic. In the extreme case where all individuals in a category (and only them) were included in a category of another variable — as was the case with Quebec physicians whose religion is Judaism and who all said their ethnic origin was Jewish — a problem of colinearity arose that would imply the impossibility of including the two variables in question in the same analysis. This is why we have set aside the results relating to ethnic origin in Quebec: this variable, which had a sizable influence on the acceptability of abortion, could not be included at the same time as religion. In the less extreme cases of correlated predictors and subcategories, adjusted deviations can differ greatly from non-adjusted deviations. Where this occurred, we indicated it by an asterisk next to the adjusted deviation.
- 35. It should be noted, however, that this distribution is not entirely normal, although it is fairly close. Variance analysis is highly resistant to slight deviations from the norm, and its reliability does not suffer.
- 36. For the sake of brevity, "expanded access to PND" is used as a synonym for "expanded access to amniocentesis and chorionic villus sampling" in the text.
- 37. "Favourable" refers to the percentage of doctors who did not answer "never" when asked when they would use predisposition tests.
- 38. (1) To support multidisciplinary teams that will provide care to socially disadvantaged pregnant women. (2) To increase the budget and improve the service of cytogenetic laboratories. (3) To improve training in obstetrical ultrasonography and increase the number of specialists doing scans. (4) To implement population-wide prenatal screening blood tests (e.g., maternal serum AFP, human chorionic gonadotropin [hCG], estriol tests) to identify women at genetic risk (trisomy 21, spina bifida), irrespective of their age. (5) To develop integrated nutritional assistance and counselling programs for women at risk in order to reduce the number of babies with low birth rate. (6) To develop wide-scale information

programs about the harmful effects of alcohol and smoking during pregnancy. (7) To develop services for the treatment of infertility.

- 39. A cleavage was considered substantial where a factor had an influence in at least three provinces, or else in two provinces and in Canada as a whole.
- 40. The general principle of group analysis is to group objects based on similarities found for a set of characteristics. A similarity index is first calculated for each pair or groups of objects already formed, then the two most similar objects or groups were combined. Beginning with the most similar objects, this process is repeated a number of times until only one group is left. The last groups formed before the end of the process are those with the highest internal homogeneity and the highest external heterogeneity.
- 41. This was the average answer concerning the timing of genetic predisposition testing (*in utero*, at birth, in adulthood, never) for five conditions (diabetes, alcoholism, schizophrenia, Alzheimer's disease, and coronary heart disease).
- 42. This was the average ranking given to the funding of medical technology, less the average ranking given to funding preventive health programs.

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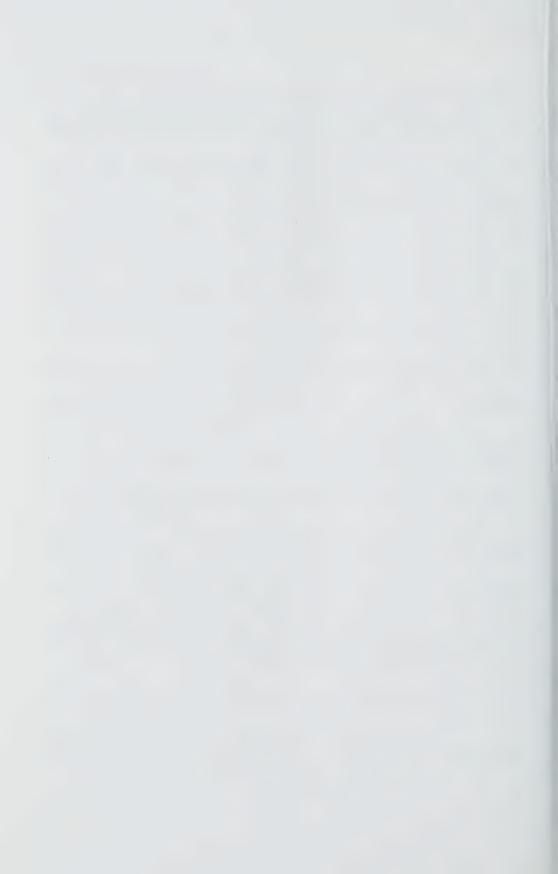
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An Analysis of Temporal and Regional Trends in the Use of Prenatal Ultrasonography

G.M. Anderson



Executive Summary

Prenatal ultrasound examination is a technology that can be used either as part of a diagnostic work-up for women suspected of having specific conditions during pregnancy or as a screening test routinely applied to women in order to help identify unsuspected conditions that could be treated, leading to an improved pregnancy outcome. Although there is agreement on the value of ultrasound examination in diagnosis, there is debate over the appropriate use of this technology as a routine screening procedure for all pregnant women.

This study used physician billing data from Ontario and British Columbia to examine the temporal trends and patterns of prenatal ultrasound use. The analysis indicated that the rate of ultrasound use doubled in both provinces between 1981-82 and 1989-1990. It was estimated that in Ontario in 1989-1990 an average of almost 2.2 prenatal ultrasound examinations were performed per delivery. In British Columbia the rate was about 20 percent lower than in Ontario. Ultrasound rates were highest in the youngest and oldest age groups. Analysis of data from Ontario indicated that the increased use of prenatal ultrasound was the result of increased provision of the service in physicians' offices rather than increased provision of the service in hospital settings. Analysis of a linked data set from British Columbia

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indicated that about 85 percent of the women who gave birth in that province had at least one prenatal ultrasound and that about 8 percent of women received more than four prenatal ultrasounds.

The overall rates of use of prenatal ultrasound in British Columbia and Ontario are higher than would be expected if ultrasound were used only for diagnostic purposes. The overall rates are consistent with rates that would be expected with a policy of routine screening of all pregnant women, but the existing pattern of use, which involves a substantial proportion of women not receiving any prenatal ultrasounds and an increasing reliance on ultrasounds late in pregnancy, is not consistent with tested screening programs. There is a need to develop acceptable guidelines for the appropriate use of prenatal ultrasound and a program to ensure that those guidelines are implemented.

Introduction

Ultrasound examination involves the transmission of high-frequency sound waves through tissue and the collection and display of the echoes produced by these waves. In the early 1950s the first crude studies of the use of ultrasound in prenatal care were undertaken. Since that time, prenatal ultrasound examination has become a highly developed technology capable of detecting many structural and functional fetal abnormalities.

Ultrasound examination can play two potentially important roles in prenatal care. The first of these is as part of the diagnostic process used for pregnancies in which there is a clinical indication of the need for an investigation. In this role, ultrasonography can be used directly as a diagnostic procedure to investigate symptoms, or indirectly as an adjunct to other diagnostic procedures such as amniocentesis and fetoscopy.

The second potential role for prenatal ultrasound is as a screening test. Screening tests are applied to asymptomatic populations in order to separate those who are at high risk for particular problems from those at low risk for such problems. The high-risk individuals are then subjected to more detailed investigation and treatment if necessary. Prenatal ultrasound screening involves examination of pregnant women for whom there is no clinical indication for the use of a diagnostic ultrasound examination to permit detection and management of potential problems. The diagnostic importance of prenatal ultrasound has been acknowledged in extensive reviews. The role of prenatal ultrasound as a routine screening test, however, has been the focus of debate.

Prenatal ultrasound screening has been suggested for various purposes including the estimation of gestational age, the detection of multiple pregnancies, the detection of fetal anomalies, placental localization, and the identification of intrauterine growth retardation. Reports from the Canadian Task Force on High Risk Pregnancies and Prenatal Record Systems, the Perinatal Medicine Committee of the Society

of Obstetricians and Gynaecologists of Canada,³ and a National Institutes of Health Consensus Conference⁴ were not in favour of the use of routine prenatal ultrasound screening. But a Norwegian Consensus Conference produced a report that was supportive of routine screening.⁵ This debate reflects, in part, the lack of clear evidence regarding the impact of prenatal ultrasound screening on maternal and fetal outcomes.

The impact of prenatal ultrasound screening was examined in seven randomized controlled trials. Three of these trials investigated the impact of serial ultrasound screening involving one examination in the second trimester and another in the third trimester. None of these trials showed a statistically significant consequence of screening on perinatal mortality or on Apgar scores. Owing to small sample sizes, however, none of these studies had the power to detect potentially important effects. A meta-analysis that combined the results from these three trials also failed to demonstrate a statistically significant impact on perinatal mortality or morbidity.

The four remaining trials examined the impact of a single ultrasound screening examination during the second trimester. Two of these trials did not reveal a statistically significant effect of ultrasound exposure on birthweight, perinatal morbidity, or Apgar scores. But both of these trials had small sample sizes and therefore did not have the power to detect potentially important impacts of screening.

One trial conducted in Sweden had a sample size of 4 997 women who had no clinical indications for ultrasound testing at 12 weeks' gestation. The results of this trial showed a statistically significant smaller proportion of low-birthweight infants (i.e., below 2 500 g) as well as a statistically significant higher mean birthweight in the screened group. There was no statistically significant effect of screening on either perinatal mortality or Apgar scores.⁹

The most recent trial involved 9 310 women. ¹⁰ The sampling process used in this study did not involve exclusion criteria, and the sample consisted of 95 percent of all pregnant women in the Helsinki area during a 19-month period. This trial showed no statistically significant impact of screening on mean birthweight, the proportion of low-birthweight infants, or Apgar scores. Perinatal mortality was significantly lower in the screened group, but 11 pregnancies were terminated before 25 weeks' gestation because fetal anomalies were detected during the ultrasound examinations. In the control group, no woman underwent an induced abortion after an ultrasound finding of congenital malformation. If the terminated pregnancies are added to perinatal deaths there is no statistically significant difference between the screened and control groups in terms of fetal survival.

Recently, the *Oxford Database of Perinatal Trials* presented a metaanalysis of the randomized trials of prenatal ultrasound screening.¹¹ The review concluded that prenatal ultrasound screening reduced the rate of induction for apparent post-term pregnancy (presumably owing to better estimation of gestational age) and that twin pregnancies were detected earlier. However, neither of these effects led to improved fetal outcomes such as higher Apgar scores or lower perinatal mortality.

To date, the clinical trials have focussed on clinical outcomes such as mortality and morbidity. It is, however, important to note that there are other potential consequences associated with screening. For example, one positive effect could be the assurance provided to parents by screening. ¹² Alternatively, screening may result in false positives, and the anxiety and stress associated with the investigation of these false-positive screening tests could have a negative impact on parents. In the Finnish trial, for example, 10 of the 30 suspected fetal abnormalities detected in the screened group disappeared in the follow-up examination. ¹³ Clearly, these and other non-clinical outcomes deserve further study.

Along with an understanding of the outcomes of screening, assessment of the appropriate use of screening must also deal with its costs. Limited resources are available for the delivery of health care services, and it is essential that those resources be used wisely.

Practitioners, patients, and governments are faced with the difficult task of deciding how to best use the technology in the context of an ongoing debate regarding the use of prenatal ultrasound screening and the lack of definitive evidence on the benefits and costs of such screening. The present study was proposed to describe temporal trends in the use and costs of prenatal ultrasound in Canada. More specifically, the objectives were to estimate as accurately as possible (1) trends in overall use and costs of prenatal ultrasound; (2) rates of use of prenatal ultrasound, including age-specific rates; (3) the distribution of use within the population of pregnant women (i.e., the proportion of women receiving zero, one, two, three, or more ultrasound examinations); and (4) differences in use across different jurisdictions in Canada. Another purpose was to determine the relative impact of changes in population size, population composition, and rates of use per capita on trends in the use of prenatal ultrasound.

The study examined trends in utilization of prenatal ultrasound in jurisdictions that have taken different approaches to access. Until 1991, in Ontario, obstetrical ultrasound examinations performed in unlicensed facilities (including both hospitals and private offices) could be billed to the provincial health care system. In British Columbia, obstetrical ultrasounds can be billed to the provincial health care plan only if they were performed in licensed facilities. These licensed facilities are almost exclusively hospitals.

Method

Data Sources

One of the advantages of Canada's universal, publicly administered health care insurance system is the generation of administrative data on

the use of health care services. The value of these administrative data in studying the use of prenatal ultrasound depends on (1) the specificity of coding for prenatal ultrasound examinations in provincial fee schedules, (2) the comprehensiveness of billing data, and (3) the availability of the billing data. A feasibility study was undertaken to assess the value of administrative data on prenatal ultrasound in relation to these factors.

The results of the feasibility study indicated that the medical service fee schedules in six provinces used specific codes for prenatal ultrasonography. In three of these provinces the information collected under these fee schedule codes was not comprehensive because a substantial proportion of prenatal ultrasound examinations involved reimbursement through hospital budgets rather than through billings for medical services.

For one of the three provinces with specific and comprehensive data, it was not possible to gain access to the data. As a consequence, administrative data from two provinces, Ontario and British Columbia, were used

in this study.

Three different sources of billing data were available. The first of these was aggregate data on the annual number and cost of specific fee code items maintained by Health and Welfare Canada (HWC). The data from HWC are aggregated at the level of billings submitted by individual physicians and can provide information on overall levels of use and costs. (Because of the level of aggregation they cannot be used to examine use across different patient age groups.) After obtaining permission from the ministries of health in Ontario and British Columbia, HWC provided access to annual aggregate data related to ultrasound use for the period 1981-82 through 1989-1990.

The second source was the provincial administrative files containing all of the individual claims submitted for services provided. Each claim contains information on the fee item, the patient's age, and whether the service was provided in a hospital or a non-hospital setting. These files are very large and, therefore, comprehensive analysis can be quite expensive. In order to control costs while providing specific information, it was initially decided to limit the analysis of these individual claims files to the fiscal vears 1981-82, 1983-84, 1985-86, 1987-88, and 1989-1990. Subsequently it was learned that, although the Ontario Ministry of Health had archived data-tapes for these periods, several tapes were not readable. It was not possible to obtain complete data for any month for 1981-82, and for 1983-84 and 1985-86 only certain months had complete sets of readable tapes. June was the only month for which complete data were available for both 1983-84 and 1985-86. Therefore, the Ontario patient-level analysis for these two fiscal years was based on data for the month of June rather than annual data. Complete annual data were available from Ontario for 1987-88 and 1989-1990 and from British Columbia for 1981-82, 1983-84. 1985-86, 1987-88, and 1989-1990.

The final data source was a data base created by the Health Information Development Unit (HIDU) at the University of British Columbia. The HIDU has developed sophisticated linkage techniques that can be used

to accurately assign services to specific patients. For this study, a data base was created that linked prenatal ultrasound scans to individual women who gave birth in British Columbia during 1986-87, 1987-88, and 1988-89.

Data Analysis

Overall Annual Trends, Rates of Use, and Costs

The Health Information Division of HWC was able to provide aggregate annual data on the number and costs for individual fee code items. In consultation with HWC, the fee codes for prenatal ultrasound in British Columbia and Ontario from 1981-82 through 1989-1990 were identified. Tables listing the total number of billed services for each of these fee code items and the total payment for these fee code items were produced by HWC.

Trends in utilization of services are the result of the impact of changes in the size of the population at risk, changes in the age distribution within that population, and changes in per capita utilization within that population. In order to isolate the effects of population size and age distribution, it was necessary to calculate per capita rates of use for prenatal ultrasound. Ideally, the denominator for these rates would be the number of women at risk for receiving prenatal ultrasound examinations and the numerator would be the sum of these examinations received by each of these women.

The population at risk for prenatal ultrasound — pregnant women — could be divided into two groups on the basis of the pregnancy outcome. The first group included women for whom the pregnancy terminated with a delivery and the second, women for whom the pregnancy terminated with an abortion. Within the group of women who aborted were two subgroups: those who had spontaneous abortions, and those who had therapeutic abortions. It was possible to use administrative billing data to determine the number of deliveries and the number of therapeutic abortions. Unfortunately, in neither British Columbia nor Ontario was it possible to assess accurately the number of spontaneous abortions from the billing data.

The relevance of determining the number of spontaneous abortions for the calculation of the rates of use of prenatal ultrasound was that ultrasound examination may be part of the diagnostic investigation of a woman suspected of having a spontaneous abortion. These women, therefore, form part of the population at risk for receiving prenatal ultrasounds. Therapeutic abortions may be less relevant to the calculation of rates of ultrasound use because ultrasound examination would not likely be part of the normal work-up prior to a therapeutic abortion; therefore, these women are not likely to be part of the population at risk for prenatal ultrasound.

Rates of use for prenatal ultrasound were calculated from the HWC data by dividing the total number of prenatal ultrasound examinations

provided in a year by the total number of deliveries during that year. Given the lack of specific data on prenatal ultrasonography associated with spontaneous abortion, these rates will tend to provide an overestimate of the true rate of use of prenatal ultrasound. The size of this overestimate will be determined by the number of spontaneous abortions, the proportion of those that involve ultrasound, and the proportion of those that are billed as prenatal ultrasound examinations. It is estimated that about 15 percent of pregnancies end in spontaneous abortions that are apparent clinically. If each of these spontaneous abortions was associated with an ultrasound, that ultrasound would be billed under a prenatal ultrasound fee code; therefore, the overall rates of use, calculated as prenatal ultrasound examinations divided by deliveries, could be up to 15 percent too high.

Age-Specific Rates of Use

Utilization of prenatal ultrasound may be determined not only by the size of the population at risk, but also by changes in the age distributions within that population. If rates of use are not equal across age categories, then shifting age structure of a population can have an impact on use rates.

The data maintained by HWC did not have information on patient age. Therefore, age-specific rates of use had to be calculated from provincial data sources. The process of calculating age-specific rates was the same as that used for the overall rates. The number of ultrasound examinations received by a specific age category during a defined time period was divided by the number of deliveries for that age category during the same time period.

Use of the Linked Data Base

The data from HWC and the administrative data from the provincial ministries of health were kept in files containing all claims submitted during a fiscal year. A potentially important methodological issue was involved in the calculation of rates of prenatal ultrasound using these aggregate data on annual use. The problem was related to the temporal relationship between prenatal ultrasounds and later deliveries. deliveries in any given year would have been associated with ultrasound examinations administered up to nine months earlier. Similarly, ultrasound examinations during any given period would be associated with deliveries up to nine months after that period. A rate defined as the number of ultrasound examinations in a jurisdiction during a year divided by the number of deliveries in that jurisdiction during the same time period would not reflect the true incidence of ultrasound use unless the true rates for both deliveries and ultrasounds were stable, in which case these two effects would cancel each other out. However, the rates were not stable and it was important to estimate the bias involved in using aggregate annual data to calculate rates.

This potential problem with the use of aggregate data and the previously noted problem related to the use of ultrasound examinations for

women who abort indicate the need for an analysis to link prenatal ultrasound examinations to specific deliveries. In theory, it should be possible to use billing data to link each woman identified as having given birth with the prenatal ultrasound services she has received. However, direct data linkage at the individual patient level using large administrative data sets such as provincial hospital discharge abstracts and medical services files is complex and resource-intensive.

Although there has been little formal experience with data linkage in Ontario, the HIDU has developed and applied linkage techniques to hospital and medical service data. More specifically, the HIDU has been involved in a study of prenatal care that provides a solid methodological framework for linking data on women who have had babies to data on their received prenatal services.

The HIDU project used hospital discharge data files to identify all women who gave birth in hospitals in British Columbia during the three-year period 1986-87 through 1988-89. Data on prenatal visits billed to the provincial medical plan were then linked to these women, using various identifiers available on the hospital discharge and medical service data files. The most powerful identifier was the Medical Services Plan (MSP) number. The hospital records for which it was possible to link the MSP number on the hospital discharge record with at least one prenatal service billed under the MSP program were defined as cases with the best hospital record because this linkage provided some indication that the MSP number on the hospital discharge abstract was accurate. Rates and frequency distributions for prenatal ultrasounds were determined using both the best hospital record file and the file containing all hospital deliveries.

The linkage process attempts to link as many of the prenatal ultrasounds as possible to specific deliveries. However, because of miscoding of MSP numbers and other linkage identifiers, the linkage process is not always able to connect all services. Rates calculated from linked data bases will tend to underestimate true incidence. The extent of that underestimate is difficult to determine accurately. Using the best hospital records, it was possible to link all but 6 percent of prenatal ultrasounds provided after 20 weeks of gestation in 1987-88 to deliveries. This suggested that the ultrasound use rates calculated from the best hospital records would underestimate true rates by no more than 5 or 6 percent. Because the records without accurate MSP numbers were less likely to be successfully linked, the analysis based on all of the identified hospital deliveries would be expected to result in less accurate estimates of true rates.

Results

Table 1 presents data on the growth in both the number and costs of prenatal ultrasound examinations in British Columbia and Ontario during

the nine-year period from 1981-82 through 1989-1990. In both provinces, there was over a twofold increase in the number of prenatal ultrasound billings. In British Columbia, the doubling in the number of prenatal ultrasound examinations was matched by a doubling of the expenditure on these services. In Ontario, increases in the fees for ultrasound procedures and shifts in the mix of fee code items billed combined with the increase in the number of services provided to produce a fourfold increase in ultrasound expenditures during the decade.

Table 1. Aggregate Data on the Number and Cost of Prenatal Ultrasound Examinations Performed in Ontario and British Columbia, 1981-82 Through 1989-1990

		Ontario		
Year	Number of ultrasounds	(% of 1981-82)	Cost in \$ thousands	(% of 1981-82)
1981-82	128 944	(100)	5 654	(100)
1982-83	146 744	(114)	7 335	(130)
1983-84	165 025	(128)	9 199	(163)
1984-85	167 731	(130)	10 162	(180)
1985-86	189 277	(147)	11 804	(209)
1986-87	218 599	(169)	14 494	(256)
1987-88	248 128	(192)	17 703	(313)
1988-89	278 656	(216)	20 314	(359)
1989-1990	312 289	(242)	22 865	(404)

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Year	Number of ultrasounds	(% of 1981-82)	Cost in \$ thousands	(% of 1981-82)
1981-82	35 822	(100)	2 561	(100)
1982-83	44 055	(123)	3 674	(143)
1983-84	49 074	(137)	4 346	(170)
1984-85	54 355	(152)	4 020	(157)
1985-86	54 181	(151)	3 823	(149)
1986-87	58 956	(165)	4 142	(161)
1987-88	64 226	(179)	4 523	(177)
1988-89	72 856	(203)	5 202	(203)
1989-1990	81 376	(227)	5 976	(233)

Source: Health and Welfare Canada.

Table 2 presents data on the rates of use of prenatal ultrasound calculated as the total number of services provided annually divided by the annual number of deliveries. Rates of prenatal ultrasound use were consistently higher in Ontario than in British Columbia. Although the rates were different in these jurisdictions, the two provinces showed similar patterns of growth in use, with rates doubling during the nine-year study period.

Table 3 presents data on age-specific rates of use. Ontario data were available only for four periods and the Ontario rates for 1983-84 and 1985-86 were based on data from a single month for each year. In both jurisdictions, the results indicated a consistent U-shaped relationship between age and ultrasound usage; use was highest for the youngest and oldest age groups and lowest for those who were 25-29 or 30-34 years of age. In both jurisdictions, use rates increased in each age group during the study period. Although the absolute growth in use rates varied across age categories, the percentage of growth was similar for all of the age categories.

It was possible to use the Ontario billing data to determine whether ultrasound services were provided in hospital or non-hospital (e.g., private office) settings. Table 4 presents data on the proportion of ultrasound examinations performed in each type of setting. The data for 1983-84 and 1985-86 were derived from billings for a single month in each year. The numbers indicate that, although the majority of ultrasound examinations were provided in hospital settings in 1983-84, by 1987-88 the majority were provided in non-hospital settings. This shift in the service setting continued into 1989-1990. Combining the data on the overall number of ultrasound services performed in Ontario with figures on the setting of

Table 2. Utilization Rates for Prenatal Ultrasound Examinations in Ontario and British Columbia, 1981-82 Through 1989-1990

	Ontario		British Col	umbia
Year	Rate per 1 000 deliveries	(% of 1981-82)	Rate per 1 000 deliveries	(% of 1981-82)
1981-82	1 059	(100)	877	(100)
1982-83	1 204	(114)	998	(114)
1983-84	1 337	(126)	1 037	(118)
1984-85	1 321	(125)	1 177	(134)
1985-86	1 474	(139)	1 251	(143)
1986-87	1 689	(159)	1 370	(156)
1987-88	1 899	(179)	1 520	(173)
1988-89	2 059	(194)	1 730	(197)
1989-1990	2 188	(207)	1 750	(200)

Source: Health and Welfare Canada.

Table 3. Age-Specific Ultrasound Utilization Rates for Ontario and British Columbia (per 1 000 Deliveries)

		Ont	tario			
		Years	of age			
Year	< 20	20-24	25-29	30-34	35-39	≥ 40
1983-84	1 637	1 328	1 186	1 140	1 471	2 717
1985-86	1 424	1 446	1 184	1 252	1 666	2 936
1987-88	2 915	1 886	1 731	1 771	2 296	4 141
1989-1990	2 857	2 247	2 028	2 006	2 405	4 615
(1989-1990 as % of 1983-84)	(174)	(169)	(171)	(176)	(163)	(170)
		British (Columbia			
		Years	of age			
Year	< 20	20-24	25-29	30-34	35-39	≥ 40
1981-82	1 045	844	839	885	945	1 260
1983-84	1 283	965	991	846	1 408	1 909
1985-86	1 623	1 220	1 152	1 180	1 626	2 344
1987-88	1 847	1 510	1 455	1 465	1 703	4 944
1989-1990	1 988	1 710	1 643	1 660	2 205	2 618
(1989-1990 as						

Source: Ontario Ministry of Health and British Columbia Ministry of Health.

(196)

(188)

(233)

(208)

(203)

(190)

Table 4. Distribution of Prenatal Ultrasound Examinations in Ontario by Place of Service

Year	% performed in hospital	% performed in non- hospital setting
1983-84	62.9	37.1
1985-86	52.3	47.7
1987-88	45.7	54.3
1989-1990	39.0	61.0

Source: Ontario Ministry of Health.

% of 1981-82)

service indicates that the number of billings for ultrasound in hospital settings increased by about 16 percent between 1983-84 and 1989-1990, while the number performed in non-hospital settings more than tripled.

The linked data base developed by the HIDU made it possible to examine not only the overall rates of use of prenatal ultrasound but also the number of exposures per person. Tables 5 and 5a present data on numbers of exposures for 1986-87, 1987-88, and 1988-89. In each year the most frequent category of use was one prenatal ultrasound procedure. In both the sample of best hospital records and the sample of all hospital records, the largest decrease in frequency occurred for women receiving no prenatal ultrasounds and the largest increase in frequency occurred for those receiving two ultrasounds. The sample of best hospital records indicated that by 1988-89 almost 85 percent of women who gave birth in British Columbia had been exposed to at least one prenatal ultrasound examination.

Table 6 presents information on the frequency distribution of prenatal ultrasound use by age group. The proportion of women not exposed to prenatal ultrasound was quite similar for the four youngest age groups. Fewer older women were not exposed to ultrasound. Those women in the two older categories were more likely to receive five or more ultrasound examinations than younger women. Over the three-year period, however, each age category studied showed an increase in the percentage of women exposed to at least one prenatal ultrasound examination.

Table 5. Frequency Distribution for Prenatal Ultrasound Examinations in British Columbia*

Number of examinations	1986-87 % of deliveries	1987-88 % of deliveries	1988-89 % of deliveries	Difference 1988-89 1986-87
0	22.1	18.5	15.3	-6.8
1.	42.2	43.3	42.6	+0.4
2	22.0	23.0	25.3	+3.3
3	8.7	8.6	10.2	+1.5
4	2.9	3.8	4.1	+1.2
5+	2.1	2.9	2.5	+0.4
	100.0	100.1	100.0	0.0

^{*} Using best hospital records.

Source: HIDU, University of British Columbia; and British Columbia Ministry of Health.

Table 5a. Frequency Distribution for Prenatal Ultrasound Examinations in British Columbia*

Number of examinations	1986-87 % of deliveries	1987-88 % of deliveries	1988-89 % of deliveries	Difference 1988-89 1986-87
0	25.8	22.4	19.6	-6.2
1	40.3	41.3	40.6	+0.3
2	20.9	21.8	24.0	+3.1
3	8.2	8.1	9.6	+1.4
4	2.8	3.6	3.8	+1.0
5+	2.0	2.7	2.4	+0.4
	100.0	99.9	100.0	0.0

^{*} Using all hospital records.

Source: HIDU, University of British Columbia; and British Columbia Ministry of Health.

Table 6. Age-Specific Distribution of Prenatal Ultrasound Examinations in British Columbia Using Best Hospital Records

Years of age	Number of examinations	1986-87 % of deliveries	1987-88 % of deliveries	1988-89 % of deliveries
< 20	0	23.3	20.7	15.8
	1	49.2	49.9	50.3
	2	20.5	20.6	24.1
	3	5.3	6.1	7.5
	4	0.9	2.2	1.7
	5+	0.9	0.6	0.7
20-24	0	24.2	19.9	16.4
	1	44.3	46.4	46.9
	2	20.7	22.8	24.2
	3	7.0	7.3	8.2
	4	2.5	2.4	2.9
	5+	1.3	1.3	1.3

Table 6. (cont'd)

Years of age	Number of examinations	1986-87 % of deliveries	1987-88 % of deliveries	1988-89 % of deliveries
25-29	0	22.8	19.4	16.2
	1	43.9	45.1	44.1
	2	20.8	22.8	25.6
	3	8.1	8.1	9.1
	4	2.6	2.8	3.2
	5+	1.9	1.9	1.8
30-34	0	22.3	18.5	15.9
	1	41.5	43.5	42.4
	2	21.6	23.8	25.0
	3	9.1	8.8	10.1
	4	3.2	3.2	4.1
	5+	2.4	2.3	2.5
35-39	0	12.9	10.9	9.2
	1	26.9	25.7	25.1
	2	33.0	23.1	28.4
	3	16.6	14.0	19.2
	4	5.6	13.3	10.6
	5+	5.0	13.0	7.5
≥ 40	0	8.4	7.6	8.0
	1	21.6	18.2	17.4
	2	33.6	25.6	23.0
	3	19.4	12.9	24.2
	4	10.3	15.3	14.8
	5+	6.8	20.3	12.7

Source: HIDU, University of British Columbia; and British Columbia Ministry of Health.

Over the period 1985-86 through 1989-1990 the British Columbia fee schedule contained two fee codes for prenatal ultrasound. One code was for ultrasound examinations performed in the early part of pregnancy and the other was for ultrasound examinations performed later in the pregnancy. Over the period 1985-86 through 1987-88 an early ultrasound was defined as an ultrasound performed prior to 20 weeks' gestation and a late ultrasound as one performed at 20 weeks' or more gestation. In 1988-89 the definitions of early and late prenatal ultrasounds were changed. An early ultrasound was defined as one performed before 14 weeks' gestation and a late ultrasound as one performed after that. These fee codes make

it possible to use the British Columbia data to examine trends in the timing of prenatal ultrasounds.

Table 7 presents data on the frequency of these two types of ultrasound examination. The numbers indicate that women were more likely to be exposed to at least one early prenatal ultrasound assessment than to at least one late prenatal ultrasound assessment. The figures also indicate that between 1986-87 and 1988-89 the likelihood that women would be exposed to at least one ultrasound of either type increased.

Table 8 provides a further description of the use of both types of ultrasound examination and their impacts on the change in total use rates. This table draws on the federal aggregate data on ultrasound use. Over the

Table 7. Frequency Distribution of Exposure to Early and Late Prenatal Ultrasound in British Columbia Using Best Hospital Records

	Number of	Early ultrasound*	Late ultrasound**
Year	Number of ear ultrasounds	% of population	% of population
1986-87	0	41.6	58.3
	1	44.5	29.4
	1 2 3 4 5+	10.6	8.7
	3	2.6	2.3
	4	0.5	0.7
	5+	0.2	0.5
1987-88	0	37.5	55.8
	1	48.3	31.5
	2	9.4	9.1
	3	2.4	2.5
	4	1.6	0.7
	0 1 2 3 4 5+	0.7	0.4
1988-89	0	35.0	51.3
	1	50.7	34.0
	0 1 2 3 4 5+	10.8	10.4
	3	2.6	3.0
	4	0.7	0.8
	5+	0.2	0.5

^{*} Prior to 20 weeks' gestation for 1986-87 and 1987-88 and prior to 14 weeks in 1988-89.

Source: HIDU, University of British Columbia; and British Columbia Ministry of Health.

^{** 20} weeks' gestation or later for 1986-87 and 1987-88 and 14 weeks or later in 1988-89.

Table 8. Fee Code-Specific Rates of Use in British Columbia

Year	Early ultrasound* rate per 1 000 deliveries	Late ultrasound** rate per 1 000 deliveries
1985-86	732	519
1986-87	821	549
1987-88	905	615
1988-89	824	906
1989-1990	563	1 187

- * Prior to 20 weeks' gestation for 1985-86 through 1987-88 and prior to 14 weeks for 1988-89 and 1989-1990.
- ** 20 weeks' gestation or later for 1985-86 through 1987-88 and 14 weeks or later for 1988-89 and 1989-1990.

Source: HIDU, University of British Columbia; and British Columbia Ministry of Health.

period 1985-86 through 1987-88, when the cut-off between early and late ultrasounds was defined at 20 weeks' gestation, the rates of both early and late ultrasounds increased. Between 1987-88 and 1988-89 the cut-off for defining an early prenatal ultrasound was dropped back to 14 weeks' gestation from 20 weeks. As expected, this fee code change resulted in a decrease in early ultrasounds and an increase in late ultrasounds between 1987-88 and 1988-89. There was a further decrease in early prenatal ultrasound rates between 1988-89 and 1989-1990. This decrease occurred during a period when the definition of an early prenatal ultrasound remained at 14 weeks. Over this same time period the rate of late prenatal ultrasounds continued to increase.

Table 9 presents data on trends in exposure to different patterns of prenatal ultrasound use. Women in the linked data set were divided into four categories determined by their patterns of exposure: (1) women not exposed to any prenatal ultrasound examination; (2) women exposed to at least one early prenatal examination, but who received no late prenatal ultrasound assessment; (3) women exposed to at least one late but no early prenatal ultrasound examination; and (4) women exposed to both early and late prenatal ultrasound assessments.

In 1986-87, 41.6 percent of women did not receive any prenatal ultrasound examinations prior to 20 weeks' gestation. This group was made up almost equally of women not having any prenatal ultrasounds (22.1 percent) and women having no early ultrasounds but at least one late

ultrasound (19.5 percent). In 1988-89, when an early prenatal ultrasound was defined as occurring prior to 14 weeks' gestation, 34.8 percent of women had no early prenatal ultrasounds.

Table 9. Frequency Distribution of Exposure to Different Patterns of Care in British Columbia Using Best Hospital Records

	Pattern of care			
Year	No prenatal ultrasounds % of deliveries	At least one early but no late ultrasounds % of deliveries	At least one late but no early ultrasounds % of deliveries	Both early and late ultrasounds % of deliveries
1986-87*	22.1	36.2	19.5	22.2
1988-89**	15.3	35.9	19.5	29.1

^{*} Early ultrasound prior to 20 weeks' gestation and late ultrasound 20 weeks or more.

Source: HIDU, University of British Columbia; and British Columbia Ministry of Health.

Discussion

Although the use of administrative billing data had advantages in terms of availability and comprehensiveness, there were some important limitations in their use to describe and analyze trends in ultrasound utilization. These limitations included the inability to separate diagnostic from screening ultrasound examinations, the failure to account for out-of-province use, the question of representativeness of data from only two provinces, and the accuracy of the estimated rates of use.

Because administrative data sets lack clinical detail on patients, it was impossible to separate ultrasound assessments used either directly or indirectly for diagnostic purposes from ultrasound examinations provided for screening purposes. The potential indications for diagnostic prenatal ultrasound are broad, and many women may require such services. There is, however, little information on either the relevant indications for diagnostic ultrasound or the incidence of such indications. However, in one recent trial of prenatal ultrasound screening, 14 about 30 percent of women

^{**} Early ultrasound prior to 14 weeks' gestation and late ultrasound 14 weeks or more.

were excluded from the trial because they had indications that warranted investigation by ultrasound.

Another potential limitation of the present study was that out-of-province use was not measured. Although out-of-province use may be an important issue for tertiary services such as coronary artery bypass surgery, it may not be as important for services such as deliveries and prenatal ultrasound examinations — services for which women are unlikely to have to travel long distances. Out-of-province use of basic obstetrical services is likely to be most relevant to remote rural areas that border on other provinces. This may be an important issue for individuals in those communities, but, given the small number of such communities, it is unlikely to have a large impact on provincial rates.

Since specific and comprehensive data on prenatal ultrasound use were available only for two provinces, this study cannot provide an accurate picture of prenatal ultrasound use in Canada as a whole. Comparable analyses for other provinces would involve either expensive primary data collection or the identification of alternative secondary data sources. Although the study was limited to two provinces, it is important to remember that these two provinces account for about 40 percent of the total Canadian population and that analysis of data from these two provinces describes a substantial component of the prenatal care.

The use of aggregate annual data on ultrasound examinations and deliveries to estimate rates of use has some important limitations. The major potential problem in this study was the overestimation of true rates owing to inclusion of prenatal ultrasound examinations that were not associated with subsequent deliveries in the numerator and then the exclusion of these cases from the denominator (i.e., ultrasounds provided to women whose pregnancies resulted in abortions). Although it was not possible to estimate the actual size of this bias, it seemed unlikely that it would inflate the rates by more than 15 percent (about 15 percent of pregnancies end in apparent spontaneous abortions that might be investigated using ultrasonography).

The linked data set was subject to a potential bias that could have led to an underestimate of true rates, since not all prenatal ultrasound examinations could be correctly linked to deliveries, because of errors in the coding of linkage identifiers. Analysis of the sample of the best hospital records indicated that this bias was unlikely to be larger than 6 percent.

One conclusion that follows is that the true absolute rates of prenatal ultrasound use likely lie somewhere between the rates calculated from aggregate annual data and those calculated from the linked data base. Moreover, if such biases remain constant over time, an accurate measure of trends in use can be provided and the determinants of those trends can be examined.

In spite of the limitations, therefore, this study may help to shed light on the use of prenatal ultrasonography. In particular, it was possible not only to describe overall trends in the use of prenatal ultrasound, but to determine the extent to which those trends were driven by changes in the basic demographic forces related to the numbers and ages of pregnant women as compared to changes in per capita rates of use. Changes in per capita use indicate either changes in the health needs of women or changes in the attitudes of providers toward the use of prenatal ultrasound. Although it may be difficult to alter demographic forces, both provider attitudes and women's health needs may be altered through policy initiatives.

It is clear from this study that there was a rapid and relatively steady increase in the number of prenatal ultrasound examinations performed in Ontario and British Columbia during the 1980s. In British Columbia, the average price for a prenatal ultrasound examination remained stable during this period, and expenditures rose in proportion to volume. This price stability in British Columbia was a reflection of the fees negotiated by the provincial medical association. In 1981 there was one fee item for prenatal ultrasound (fee code 8651) with a fee of \$74.90. In 1989-1990 there were two fee codes for prenatal ultrasound. One (fee code 8651) had a fee of \$76.50 and was for a scan at 14 weeks' or more gestation and the other (fee code 8655) had a fee of \$67.00 for a scan performed at less than 14 weeks' However, in Ontario the average price for an ultrasound examination increased at the same time as the rapid increase in volume. resulting in a fourfold increase in expenditures measured in nominal dollars and almost a threefold increase in expenditures deflated by the consumer price index. Again, this average price increase reflected fees negotiated by the provincial medical association. In 1981, there was one main fee code for prenatal ultrasound (fee code J159) in Ontario and the total fee for that service was \$45.90. In 1988-89 the fee for that item had increased to \$74.60.

The data from HWC indicate that there were 183 345 more ultrasound examinations performed in Ontario in 1989-1990 than in 1981-82 and 45 554 more performed in British Columbia in 1989-1990 than in 1981-82. A major goal of this paper was to determine the relative impact of demographic factors and factors related to per capita utilization rates on these increases. Conceptually, one way to look at the role of demographic and utilization rate changes in these volume increases was to start with the 1981-82 population and calculate the impact on the total volume of ultrasound examinations of changes in the size of the population, the age structure of the population, and the utilization rate.

In Ontario the number of deliveries increased by 17 percent from 1981-82 to 1989-1990; in British Columbia during the same period, the number of deliveries increased by 14 percent. The effect of this growth in the number of deliveries on the volume of services could be estimated by calculating the number of ultrasound examinations that would have been expected in 1989-1990 if both the age structure and the utilization rate had remained the same as in 1981-82, but the population had grown to 1989-1990 levels. This is simply the product of the 1981-82 overall rate (i.e., 1 059 per 1 000 deliveries in Ontario) and the 1989-1990 population

(i.e., 142 712 deliveries in Ontario). This works out to an expected 151 132 ultrasound examinations (22 188 more examinations than were observed in 1981-82). Thus, about 12 percent of the increased volume of ultrasound examinations in Ontario could be attributed directly to the increased number of deliveries. Using a similar approach it was estimated that about 11 percent of the increased volume of ultrasound procedures in British Columbia could be attributed to the increased number of deliveries.

The impact of relative changes in age in the population of pregnant women on the volume of services provided could be estimated using the age-specific rates calculated from provincial billing data. If only the age structure had changed between 1981-82 and 1989-1990, the expected number of ultrasound examinations in 1981-82 could be calculated using the 1989-1990 population distributions and the 1981-82 age-specific rates of use. British Columbia has comprehensive provincial age-specific data for both 1981-82 and 1989-1990. Applying the 1989-1990 population age distribution to the 1981-82 age-specific rates resulted in an expected overall rate of 895 per 1 000 deliveries. This expected rate times the 1981-82 population of 40 828 deliveries yielded 37 436 expected ultrasound examinations. This is 1 614 more than observed in 1981-82 and explains less than 4 percent of the total increase in the use of prenatal ultrasound. This effect was small, although there was a substantial increase in the average age of women who gave birth in 1989-1990 compared to 1981-82, because of the U-shaped relationship between age and utilization rates. The provincial age-specific data from Ontario were not available for 1981-82. Using the 1983-84 monthly data for age-specific rates and the 1989-1990 age distribution of deliveries it was estimated that changes in age could similarly account for about 4 percent of the difference in volume of prenatal ultrasounds between 1983-84 and 1989-1990 in Ontario.

In order to determine the effect of the change in utilization rates alone, the 1989-1990 age-specific rates were applied to the 1981-82 population. In British Columbia, this resulted in 70 333 expected ultrasound examinations in 1981-82, which was 34 511 more examinations than were observed in that year. Changes in utilization rates alone account for almost 76 percent of the total change in volume. Analysis of the Ontario data was confounded by the lack of age-specific data for 1981-82. However, given the small impact of age structure on volume changes, the effect of utilization can be approximated by examining the impact of differences on overall rather than age-specific rates. Application of the 1989-1990 overall rate of ultrasound utilization to the 1981-82 total population resulted in an expected 266 461 ultrasound examinations in 1981-82. This was 137 517 more than were observed, and accounts for 75 percent of the total increase in volume of ultrasound examinations in Ontario.

The independent effects of population size, population structure, and utilization rates account for about 90 percent of the increased volume of ultrasound services in both provinces. The remaining proportion of the

increase is the result of interactions among these factors, primarily the increased use rates in the larger population.

The results of these analyses indicate that the predominant reason for the increase in the volume of prenatal ultrasound examinations was an increase in per capita rates of utilization. Demographic forces alone, and their interactions with changes in use rates, account for only about onequarter of the increased volume of services. The remainder of this study focusses on the factors associated with this increase in utilization rates.

In Ontario, it was possible to define the setting in which ultrasound examinations were performed. The analysis of utilization along this dimension suggested that the increased use of services was primarily associated with the rapid expansion in the number of ultrasound examinations provided in non-hospital settings. This increase in non-hospital use of ultrasound could indicate increased access to needed services. Alternatively, the growth in non-hospital use could indicate a decrease in the threshold for ordering prenatal ultrasound — a decrease that could be associated with a physician ordering a test that could be provided and billed by that physician. Whatever the case, the provision of prenatal ultrasound examinations in unlicensed facilities in physicians' offices may play a role in explaining the higher rates of prenatal ultrasound use in Ontario compared to British Columbia. The need to license ultrasound facilities in British Columbia may allow the government to control the diffusion of ultrasound use.

The availability of a linked data base in British Columbia made it possible to examine the relationship between the increased overall rate of prenatal ultrasound and the frequency distributions of ultrasound examinations per delivery. Between 1986-87 and 1988-89 fewer women were not being exposed to prenatal ultrasound and more women received more than one prenatal ultrasound. In the period when the sample of patients with the best medical records had a 13 percent increase in the overall rate of ultrasound use, the proportion of women not exposed to prenatal ultrasound decreased by 30 percent, and the proportion of women exposed to four or more ultrasound examinations increased by 30 percent.

Although the lack of linked data for other time periods in British Columbia or for Ontario makes it impossible to directly estimate the change in the distribution of ultrasound examinations, extrapolation of the results from the linked data base suggests that the rapid increase in the use of prenatal ultrasound has been associated with both increasing rates of exposure to multiple ultrasound examinations and decreasing rates of non-exposure. By 1988-89, approximately 85 percent of women who gave birth in British Columbia had been exposed to at least one ultrasound examination and approximately 40 percent had received two or more prenatal ultrasounds. Given the higher overall rates of use in Ontario, it is likely that the number of women receiving at least one or more prenatal ultrasound examination was even higher.

The data from British Columbia also make it possible to examine the use of ultrasound provided both early and late in pregnancy. The analysis of trends in the timing of prenatal ultrasounds is complicated by the change in 1988-89 of the definition of early prenatal ultrasound from prior to 20 weeks to prior to 14 weeks of gestation. Despite these complications it appears that late prenatal ultrasound has played a larger role in explaining the higher overall use rates than early prenatal ultrasound.

The data on early and late prenatal ultrasounds can also be used to determine the patterns of exposure. The two largest trials of routine prenatal ultrasound screening employed examinations early in gestation. In the Swedish trial¹⁵ the ultrasounds were performed at 15 weeks' gestation and in the Finnish trial¹⁶ the ultrasounds were performed between 16 and 20 weeks' gestation. The 1986-87 data from British Columbia indicate that 41.6 percent of women who gave birth did not receive any prenatal ultrasounds prior to 20 weeks' gestation. The patterns of use prior to 20 weeks' gestation may have changed in more recent years in British Columbia, but the change in the definition of early and late examinations in the fee schedule makes this hard to document.

The analysis of secondary data can provide information on the level of, trends in, and determinants of prenatal ultrasound use, but it cannot provide direct evidence on the appropriateness of that use. The first step in defining appropriateness is the development of standards that define optimal care. These standards should be based on evidence derived from well-designed clinical trials.

Conclusions

Currently there is evidence from two large randomized trials¹⁷ and one recent meta-analysis¹⁸ that can provide some information on the benefits of prenatal ultrasound screening. If the examination of benefits is limited to fetal mortality and perinatal morbidity, then the current evidence does not provide overwhelming support for routine prenatal ultrasound screening. There are no statistically significant effects on Apgar scores and a significant reduction in perinatal mortality was found in only one trial, not in the meta-analysis. Moreover, the significant effect on perinatal mortality noted in the single trial was related to increased pregnancy terminations rather than increased fetal survival brought about by successful prenatal therapy.

On the other hand, prenatal ultrasound use does show significant benefits in terms of increased early detection of twins, decreased rates of induction, increased birthweight, and earlier detection of anomalies. Along with improving the evidence on fetal outcomes, there is a need for more detailed analysis of the positive and negative psychological effects of screening on parents. As a result of the lack of definitive evidence on the

benefits, there is disagreement among expert panels on the need for routine prenatal ultrasound examination.

Another step in determining the appropriateness of use requires comparison of care provided to standards for optimal care. Even if accepted standards existed, the secondary data available for this study do not have the clinical detail required to directly assess appropriateness. Studies of appropriateness will have to involve the collection of data from detailed data sources such as medical records.

Although this study cannot provide direct evidence on the appropriateness of the use of prenatal ultrasound, it can shed some light on some important clinical policy concerns. One of these concerns is that there are very high levels of prenatal ultrasound exposure in a large segment of the population. The analysis of the linked data base in British Columbia indicates that less than 7 percent of women who had babies in 1988-89 were exposed to four or more ultrasound examinations. Multiple ultrasound examinations were most commonly administered to older women. Repeated ultrasounds may be an important component of the care of certain high-risk pregnancies, in which case the current levels of multiple exposures may be justified in clinical terms.

Another important concern is the effect that the implementation of different clinical policies might have on current patterns and levels of overall utilization. If the accepted clinical policy was that routine prenatal ultrasound screening was not justified and that ultrasound should be used for diagnostic investigation of pregnancies in which there is some clinical indication, then there would likely be a large decrease in the number of ultrasound assessments performed. For example, if, as was found in the Swedish trial, ¹⁹ about 30 percent of women have clinical indications for ultrasound examination and each of these women were to receive two ultrasound examinations, then the overall rate of use would be 600 per 1 000 deliveries. This figure is far below current levels of use in either British Columbia or Ontario.

If the accepted clinical policy was that a single screening ultrasound was justified, then rates would be similar to those currently found in Ontario or British Columbia. The screening program studied in the Swedish randomized trial²⁰ resulted in an average of 1.3 ultrasounds per screened individual. If one assumes that the 30 percent of the population excluded from that trial were exposed to an average of two ultrasounds each, then the overall population rate would be about 1 500 per 1 000. In the Finnish trial,²¹ there were no defined exclusion criteria and the overall rate of use was 2 100 ultrasound examinations per 1 000 deliveries.

Implementation of routine screening programs such as those used in the two randomized trials would result in overall levels of prenatal ultrasound use that are consistent with those found in Ontario and British Columbia in 1989-1990. However, the pattern of ultrasound delivery would be different from that currently observed, in two important ways. First, routine screening would involve providing prenatal ultrasound to all

women, not only to the approximately 85-90 percent that are now receiving at least one ultrasound. Second, both of these studies involved screening ultrasound provided before 20 weeks of gestation, while the analysis of billing data shows that only about 40 percent of the women who gave birth in British Columbia in 1986-87 received an ultrasound examination by this time.

Preliminary attempts to model the impact of different clinical policy options suggest that implementing either a policy limiting prenatal ultrasound to diagnostic purposes or a policy of routine screening would result in substantial changes in current practice. There would be either a large reduction in the use of ultrasound or a substantial shift in the type of services provided.

Quality assurance involves setting standards for appropriate care, comparing current practice to those standards, and, if necessary, acting to bring practice in line with standards. Prenatal ultrasound could provide an important example for the development of such a quality-assurance process. However, the development of this process will involve a commitment to producing and interpreting research on clinical effectiveness, a willingness to systematically assess current practice, and a responsibility to act if care is inappropriate. This can occur only with the cooperation of the public, governments, and the medical profession.

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Maternal Serum AFP Screening Programs: The Manitoba Experience

B.N. Chodirker and J.A. Evans



Executive Summary

A raised level of alpha-fetoprotein (AFP) in the blood serum of pregnant women means that the fetus is at increased risk for a variety of malformations, in particular neural tube defects (anencephaly and spina bifida). It has also been recognized more recently that a decreased maternal serum AFP level is an indication of increased risk for fetal chromosomal syndromes.

While maternal serum AFP screening is now part of routine prenatal care in many parts of Europe and North America, the only provincial screening program is in Manitoba. A pilot study in 1982-83 led to the present program. This report provides an historical overview and gives data on the performance of the Manitoba Maternal Serum Alpha-Fetoprotein Screening Program, reviews the protocols used by the program, gives 1990 and some 1991 program statistics, and reviews the outcomes of the pregnancies of patients found to have abnormal maternal serum AFP values in 1990. It also reviews the impact of maternal serum AFP screening on the rates of neural tube defects in Manitoba, and provides the results of a survey of physicians' attitudes toward maternal serum AFP screening.

The Manitoba maternal serum AFP program screened 10 362 pregnancies in 1990. Of these, 389 (3.8 percent) had values that were

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considered elevated, and 441 (4.3 percent) had values that were considered low. The majority (313) of those with elevated levels had fetal assessments performed; 42 were found to have abnormal fetal assessments, including fetal death, fetal anomaly, placental anomaly, and oligohydramnios. Other reasons for the elevated levels included multiple pregnancies and incorrect dates.

Of the 441 women with low values, 105 were not considered to be at increased risk for Down syndrome. Reasons for the low values for these women included molar pregnancies, missed abortions, and overestimated gestational age. The other 337 women were at increased risk for Down syndrome because of their age combined with their low maternal serum AFP values, but some had already had prenatal testing for advanced maternal age. Of 221 who were routinely screened Manitoba patients, 138 were between 30 and 34 years of age, and thus would not normally have been offered prenatal diagnosis. Seventy-two (52.2 percent) of these women had an invasive test performed, while another 13 (9.4 percent) declined it. Of the remaining 53 women, 13 had incorrect estimates of gestational age, 37 were not referred for invasive testing, and 3 others had a molar pregnancy, fetal loss, or an incorrectly completed requisition. Of the 83 patients 35 years of age or older, 28 (33.7 percent) had an invasive test performed; 18 (21.7 percent) declined the test; 34 were not referred for invasive testing; and 3 had incorrect estimates of gestational age.

Several differences were documented between rural and Winnipeg practices with regard to prenatal diagnosis and maternal serum AFP testing. Some of the differences can be related to geographic factors and to differences in physician education and awareness. These inequalities could be reduced through increased education; prenatal outreach clinics may also be beneficial.

The report also examines the impact of the maternal serum AFP screening program on the birth frequency of neural tube defects. As is outlined in the report, this has been significant. While the overall frequency of such defects did not fall, the number of neural tube defects at birth has fallen 52 percent, due to increased prenatal detection of the defects and parents' decisions to terminate such pregnancies.

Finally, the report also provides the results of a survey of Manitoba physicians providing prenatal care about their knowledge of and attitudes toward the program. The need for more education of physicians emerges from the findings, as a significant number were uncertain about how the 35-year-old age cut-off is measured (at date of testing or due date). The survey also revealed that less than 40 percent (37.7 percent) of physicians seek patients' specific consent for maternal serum AFP testing, and that more than two in five (41.5 percent) do the test automatically either without seeking consent or unless the patient specifically declines.

Physician and patient education is also important in regard to counselling. If women were counselled before an "abnormal" result is received, anxiety would likely be less. Yet the report found that 6.6 percent of physicians do not supply any information before testing.

In general, physicians felt that maternal serum AFP was a better than average test, and most strong criticisms related to the question of pregnancy termination. Physicians were also concerned by the increased anxiety caused by a positive test, especially related to an increased risk for Down syndrome.

The Manitoba experience with maternal serum AFP indicates that such screening could be incorporated into standard prenatal practice in a coordinated and timely fashion. The impact of the program in terms of reduced birth prevalence of neural tube defects is clear, and an effect on Down syndrome is starting to become apparent. Most patients and physicians support the availability of screening, though it is important that it be offered in a way that leaves the decision whether or not to be screened to the individual patient.

Introduction

Historical Perspective

Neural tube defects, by both their nature and relative frequency, are one of the most distressing congenital malformations in humans. Of the most common types, anencephaly leads inevitably to the stillbirth or neonatal death of a severely malformed infant, while open spina bifida often leads to severe physical and, in some cases, mental handicap. The incidence of neural tube defects varies between regions, but in Manitoba about 22 infants or fetuses per year (about 1 in 775 pregnancies) have this disorder, making it one of the most common major congenital malformations.

Although the precise cause of neural tube defects is unknown, isolated defects are believed to be inherited in a multifactorial fashion influenced by genetic predisposition and environmental factors. However, as 90 percent of such infants are born to families who did not know they were at risk, the risks for an individual family cannot usually be identified until after the birth of an affected child (U.K. Collaborative Study on Alpha-Fetoprotein 1977). In addition, neural tube defects frequently occur as part of a large number of genetic syndromes and other patterns of multiple malformations (Main and Mennuti 1986). With the exception of a few chromosomal syndromes known to be associated with late maternal age, risk factors for such disorders are rarely recognized before the birth of a child with anomalies. Women with certain chronic health problems, including insulin-dependent diabetes mellitus and alcoholism, and those on specific anti-convulsant medications are also at increased risk of having affected fetuses (Friedman 1982; Main and Mennuti 1986). Precise evaluation of this risk requires investigation of the woman, her health status, and her family history.

Awareness of increased risks for certain groups of women led in the 1970s to the development of amniotic fluid alpha-fetoprotein (AFP)

determination as a test for neural tube defects (Ainbender and Hirschhorn 1976; Brock et al. 1975; Brock and Sutcliffe 1972). This test, coupled with ultrasonographic fetal assessment and quantitative and qualitative determination of acetylcholinesterase in amniotic fluid, has provided a highly sensitive tool for identification of neural tube defects prenatally. However, this testing is invasive, as it requires a sample of amniotic fluid obtained by amniocentesis, which is associated with a risk of fetal loss. Also, it cannot be offered to more than a small proportion of the women at risk of having a child with neural tube defects, because they cannot be identified in advance. It became apparent in the late 1970s that development of a safe and rapid screening process for fetal neural tube defects, which could be offered to all pregnant women, was required if many families were to be given the option of avoiding this disorder through prenatal diagnosis (U.K. Collaborative Study on Alpha-Fetoprotein 1977). Maternal serum AFP levels proved to be highly sensitive for fetal neural tube defects; they also had the unanticipated effect of identifying women at risk of having a child with Down syndrome and certain other high-risk pregnancy situations.

AFP is the major protein in the fetal circulation after albumin. Although its role in fetal development is not known, it reaches maximum concentration in fetal plasma at 12 to 15 weeks' gestation, then shows a gradual decrease to term, followed by a rapid drop in perinatal life to "adult" levels of 0 to $15~\mu g/L$ (Brock and Sutcliffe 1972). AFP is also found in amniotic fluid and, due to transplacental passage, in maternal serum. Maximum concentrations in amniotic fluid are found at 12 to 14 weeks' gestation and decrease to term. Levels in maternal serum start to rise above pre-pregnant levels at about 13 weeks' gestation, continue to rise until approximately 32 weeks' gestation, and then gradually decline. The rise in diabetic women is delayed by about 2 weeks.

Significant elevations of amniotic fluid AFP have been shown to be associated with open neural tube defects (Brock et al. 1975; Leek et al. 1973), other congenital disorders including abdominal wall defects (Kunz and Schmid 1976), Finnish-type congenital nephrosis (Kjessler et al. 1977b), blood-stained amniotic fluid (U.K. Collaborative Study on Alpha-Fetoprotein 1979), and fetal death (Seller et al. 1974). Similarly, raised maternal serum AFP levels have also been shown to be associated with the above-noted congenital malformations (Cuckle et al. 1989; Kjessler et al. 1977b; Kunz and Schmid 1976; Leek et al. 1973; Seller et al. 1973), multiple pregnancies (Johnson et al. 1990; Lidbjörk et al. 1977b; Wald et al. 1975), and other poor pregnancy outcomes, including spontaneous abortion (Lidbjörk et al. 1977b; U.K. Collaborative Study on Alpha-Fetoprotein 1979), perinatal death (Waller et al. 1991), low birthweight (Brock et al. 1977; Katz et al. 1990; Kjessler et al. 1977a; Macri et al. 1978; Wald et al. 1977), and maternal disease (Milunsky and Alpert 1978). Significantly decreased levels of maternal serum AFP have been associated with spontaneous abortion (Lidbjörk et al. 1977a, 1977c) and trophoblastic disease, especially hydatidiform mole (Kjessler et al. 1977c; Lidbjörk et al. 1977a). Thus, abnormal levels of AFP in amniotic fluid, maternal serum, or both frequently indicate potential problems in pregnancy; they are not specific for neural tube defects or any other condition. It should also be kept in mind that they are sometimes coincidental findings in what are apparently entirely normal pregnancies.

Several factors and limitations of the test have to be considered in the evaluation of maternal serum AFP screening. The multiplicity of causes of abnormal maternal serum AFP levels and the limitations in the sensitivity of the test in detecting neural tube defects mean there is the possibility of missing open defects (false negatives). The limitations in the specificity of the test mean that it may be in the abnormal range while the fetus is normal (false positives). Another important factor is the changing levels of AFP with gestational age in maternal serum and amniotic fluid and thus the need to know accurately the gestational age to correctly interpret the results. Another factor is cost, notwithstanding studies that document the potential cost-effectiveness of such screening programs (Layde et al. 1979; Sadovnick and Baird 1983). However, perhaps the most important consideration is the impact of screening on the pregnant woman and her desire to use this test or not. These concerns were recognized early by those working in this area, and an international workshop in 1979 recommended the following:

Where pilot studies have been undertaken it is accepted that maternal serum AFP screening should be available to all pregnant women who wish to participate. Sufficient information about the purpose and nature of such programmes should be available to women before pregnancy or within the first trimester so that they can make informed decisions about participation. This information can be provided by counselling in early pregnancy and at each stage of the screening and diagnostic process. (Milunsky et al. 1980, 26)

In Manitoba, as elsewhere, the local genetics community was aware of the ongoing research and development of AFP testing. Amniotic fluid AFP determinations were being offered to all women known to be at increased risk, identified because they had a child or other close relative with neural tube defects. In addition, amniotic fluid AFP levels were carried out on samples from women undergoing amniocentesis for other reasons, usually advanced maternal age. Maternal serum AFP levels for these women had also been obtained before amniocentesis to provide additional information useful in determining the precise cause of elevated amniotic fluid levels; thus, a normal range of values for maternal serum AFP at gestations between 15 and 19 weeks was already available.

Maternal serum AFP screening on a population basis was not considered in earnest until 1979 when some physicians in Winnipeg, having read of the potential of maternal serum AFP screening in the medical literature, started to screen their pregnant patients. This came to the attention of the geneticists when the first patient with an elevated value

was referred to them for further investigation and counselling. If maternal serum AFP tests are done without care being taken to ensure adequate quality control of the assay at the laboratory, without appropriate determination of the data required for correct interpretation (such as precise gestational age), without availability of resources for further investigation and follow-up of the patient, and without counselling of the women concerning the purpose of screening and the implications of an abnormal result, it is evident that the potential for harm is great. Therefore, in this situation, the genetics group sought funding for a local pilot study on the feasibility of population maternal serum AFP screening in the province. The recommendations of the 1979 international workshop were followed, and the study protocol was approved by the ethics committee of the University of Manitoba.

Funding was approved by the Manitoba Medical Services Foundation Inc. in 1981. The study team consisted of members of the departments of Human Genetics, Paediatrics, and Obstetrics at the University of Manitoba, as well as members of the Cadham Provincial Laboratory, the Immunology Laboratory at the Health Sciences Centre, and the Manitoba Medical Services Commission, and representatives from a wide range of urban and rural medical practices. The study population consisted of women referred from a variety of health care sources, including a hospital outpatient obstetric clinic, several large urban private obstetric practices, several rural general or mixed general/obstetric practices, and an urban family practice unit. All pregnant women in the participating practices seen before 18 weeks' gestation were eligible to participate. They received written material describing the study and giving details of AFP screening. All women who wished to participate had to complete a consent form. Questionnaires were given to women before screening and after they had the results of the test to assess their knowledge, to gauge anxiety, and to learn from their written comments and concerns about testing. Women who declined testing were also invited to complete the pre-test questionnaires. Log books were kept of all prenatal patients in each practice and their pregnancy outcomes where possible.

Between February 1982 and October 1983, 2 045 women were screened. A further 257 women declined testing, and in 943 cases patients were not offered testing at their 15 to 18 weeks visit. Three women in the screened population were, on further evaluation, found to have a fetus with a neural tube defect. All three elected to seek a termination of the pregnancy. No cases of neural tube defects were missed in the other screened patients, and no losses following amniocentesis occurred in the women who had this test to evaluate high maternal serum AFP levels. High response rates were received for both the pre-test (74.2 percent) and post-test (68.5 percent) questionnaires, and 98 women who declined testing (38.1 percent) also returned questionnaires. Detailed information on these

data is available (Evans et al. 1986).

In summary, screened women were supportive, and most suggested screening should be more widely available. Some women who had declined commented on the potential relation between screening and abortion, and both screened and non-screened women noted that maternal choice concerning screening should be maintained. Over 96 percent of screened women thought that they would choose to be screened in subsequent pregnancies and that it should be available to other women in Manitoba. Of the respondents who had declined screening, 35 percent thought it should be available to all and 18 percent said they would want to have it in a subsequent pregnancy. Due to the widespread support for maternal serum AFP screening in Manitoba and the availability of a central well-controlled laboratory and resources for follow-up of patients, maternal serum AFP screening continued to be available in Manitoba after the pilot study. On 1 April 1985 it became a formal provincial program funded by the Manitoba Health Services Commission.

Some changes to screening protocol since that time have been made. for example the inclusion of increased Down syndrome risks in interpretations since 1986. Observations by Merkatz et al. (1984) had suggested an association between reduced levels of maternal serum AFP and increased risk of chromosomal disorder in the fetus, especially Down syndrome. This was confirmed by other independent researchers (Cuckle et al. 1984, 1987; Hershey et al. 1986; New England Regional Genetics Group 1989) and led to the development of risk tables for Down syndrome on the basis of both age and maternal serum AFP. These provide more precise risk figures for counselling than those based on maternal age alone. Thus, women whose maternal serum AFP and age-corrected risk is greater than that of a 35-year-old (37-year-old before May 1991) are offered amniocentesis. In addition, the literature now suggests that screening incorporating additional biochemical markers such as beta-human chorionic gonadotropin (hCG) and estriol can improve Down syndrome detection (Bogart et al. 1987; Wald et al. 1988a, 1988b). We have, therefore, initiated studies to determine the normal range of data for these measurements, and we are considering the feasibility of introducing this "triple test" into the Manitoba screening program.

Since the pilot program was initiated in 1982, the proportion of prenatal patients screened by maternal serum AFP in Manitoba has increased rapidly; at present, approximately 60 percent are screened. This program is therefore the most comprehensive in Canada and is still the only province-wide program. Although the total number of patients screened is somewhat higher in the Toronto area (Wilson 1992), the Manitoba program has experience in screening women in a much wider geographical area with a variety of primary health care providers. In addition, the provincial base of the program has allowed detailed follow-up of all screened patients through the Manitoba Health Services Commission records and, using the provincial Congenital Anomalies Registry and other resources, has allowed evaluation of the impact of screening on the

prevalence in our population of neural tube defects and other pertinent conditions. Thus, the Manitoba Maternal Serum AFP Screening Program provides a unique resource with which to document the impact of maternal serum AFP screening on a Canadian population.

Study Objectives

The purpose of this study is to present data pertaining to several aspects of maternal serum AFP screening in Manitoba. The following data will be provided:

- 1. current protocols in use by the Manitoba Maternal Serum AFP Screening Program;
- 2. review of the 1990-91 program statistics using the program's computer data base files;
- 3. review of 1990 patients with abnormal maternal serum AFP values;
- 4. review of the rates of neural tube defect in Manitoba since 1979 with analysis of the impact of maternal serum AFP screening; and
- 5. results of a survey of physicians' attitudes toward maternal serum AFP screening.

Current Protocols

The Manitoba Maternal Serum AFP Screening Program is run primarily by a program coordinator and a program assistant under the supervision of one physician geneticist, one Ph.D. geneticist, and one perinatologist. The program works closely with the Fetal Assessment Unit at the Health Sciences Centre in Winnipeg, where most of the women are followed up, and with the Cadham Provincial Laboratory, where the biochemical testing is done.

A sample of the requisition filled in by physicians for the program is given in Appendix 1. Physicians are asked to supply relevant information for patient identification and determination of gestational age. The physician is also questioned about other factors that may have an impact on patient management (e.g., family history). Pamphlets describing maternal serum AFP screening are provided for physicians to give to their patients before testing (Appendix 2).

A computerized system to handle the routine interpretations was started in January 1990. As maternal serum AFP levels rise with advancing gestation, median values have been collected for each half-week

interval from 14.5 to 24.5 weeks. Given the maternal serum AFP value and the gestation on the day the sample was drawn, a MOM (multiple of the median) can be calculated. As maternal serum AFP levels are generally lower in heavier women, the MOM is also corrected for maternal weight. If the woman is diabetic during the pregnancy, her maternal serum AFP level is expected to be lower as well; to compensate, the computer subtracts two weeks from the calculated gestation for diabetic women.

The basic program protocol is shown in Figure 1. Although our recommended time for screening is 15 to 20 weeks' gestation (15 to 18 is ideal), samples can be interpreted for any gestation from 15 to 24.5 weeks. Two basic interpretations are made for each sample received at an appropriate gestation. The first interpretation is made on the basis of the MOM. A fetal assessment is suggested for either low values (less than or equal to 0.25 MOM) or high values (greater than or equal to 2.3 MOM). A dating ultrasound is recommended for borderline low values (0.3 to 0.4 MOM). No follow-up is suggested for normal values (0.45 to 2.2 MOM). In addition to the interpretation made on the basis of the MOM, an interpretation and recommendation are also made on the basis of the estimated risk for having a live-born child with Down syndrome. The risk of having a child with Down syndrome increases with advancing maternal age and at age 35 is estimated to be 1 in 384. Mothers carrying a fetus with Down syndrome tend to have low maternal serum AFP levels. Any level less than or equal to 0.8 will increase the risk of having a baby with Down syndrome over the age-related risks. Table 1, which is based on the data provided by Cuckle et al. (1987), is used by our program to determine a woman's risk of having a live-born child with Down syndrome. The maternal serum AFP level in MOMs is shown across the top, while the maternal age is shown down the left-hand column. The intersecting point of these two values is the reciprocal of the Down syndrome risk. example, a 34-year-old woman with a maternal serum AFP of 0.7 MOM has a 1 in 360 risk of having a child with Down syndrome. Blank cells on the table reflect Down syndrome risks less than 1 in 384, which are not reported by our program. During the first part of the study period, an amniocentesis was offered, based on the maternal serum AFP level, only if the risk was equivalent to at least that of a 37-year-old, and, therefore, a woman was not considered to be at increased risk unless she was at least 30 years of age. These calculations were based on the data of Hershey et al. (1986). In May 1991, this was changed, and we currently offer an amniocentesis to any women whose risk is equal to or greater than 1 in 384. Any woman 35 years of age or older is considered to have a risk for having a child with Down syndrome of at least 1 in 384 and is offered an A woman who is 34 years of age will be offered an amniocentesis if the maternal serum AFP level is 0.7 or less. A woman as young as 27 years of age may be offered an amniocentesis, but only if the maternal serum AFP level is 0.4 or less.

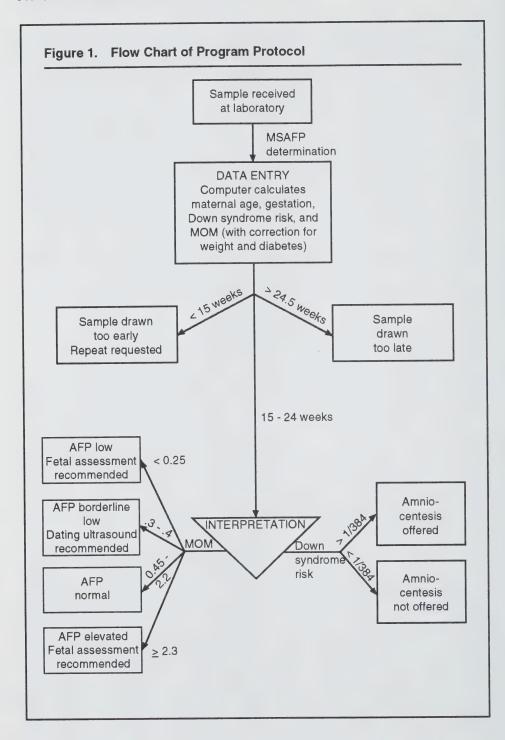


Table 1. Down Syndrome Risk Based on Age and AFP

	Maternal serum AFP level in MOMs									
Age	Not done	0.8	0.75	0.7	0.65	0.6	0.55	0.5	0.45	0.4
27										380
28										360
29										320
30									350	290
31								360	300	250
32							360	310	260	220
33						340	300	260	220	180
34				360	320	280	250	210	180	150
35	384	360	320	290	260	230	200	170	150	120
36	307	280	260	230	210	180	160	140	120	98
37	242	220	200	180	160	140	130	110	93	77
38	189	170	160	140	130	110	99	85	72	60
39	146	140	120	110	99	87	76	66	56	46
40	112	100	94	85	76	67	59	51	43	36
41	85	79	72	65	58	51	45	39	33	27
42	65	60	54	49	44	39	34	29	25	21
43	49	45	41	37	33	29	26	22	19	16
44	37	34	31	28	25	22	19	17	14	12
45	28	26	23	21	19	16	14	12	11	9
46	21	19	17	16	14	12	11	9	8	7
47	15	14	13	12	10	9	8	7	6	5
48	11	10	9	9	8	7	6	5	4	4
49	8	8	7	6	6	5	4	4	3	3

Note: Blank cells have risk less than 1 in 384.

Thus, the program does not currently decrease the number of women who are offered amniocentesis, but also offers it to younger women who are identified as being at significantly greater risk than their age alone would suggest.

A printed report is sent by mail to the physician for all patients. If the result is abnormal, either low or high, the physician's office is contacted by telephone to decrease any time delay. An example of the printed report is given as Appendix 3. Possible messages or interpretations are listed in Table 2.

Message #	Message
1	THE AFP IS BORDERLINE LOW. As the dates are confirmed and the risk for Down syndrome is below the cut-off at which an amniocentesis is offered, no further follow-up is recommended.
2	THE AFP IS LOW. We recommend a fetal assessment be done Please call 788-6240 to arrange.
3	THE AFP IS BORDERLINE LOW. Please confirm the dates with ar ultrasound (at >14 weeks' gestation) and send us a report. If the dates are accurate no further follow-up is necessary.
4	THE AFP IS LOW. We recommend a fetal assessment be done. It the gestation is accurate an amniocentesis will be offered at that time.
5	THE AFP IS NORMAL. If the gestation is considered accurate, no further screening is necessary.
6	THE AFP IS LESS THAN THE MEDIAN. The risks for Down syndrome are therefore increased. If an amniocentesis is desired call 788-6240 to arrange.
7	THE AFP IS ELEVATED. We recommend a detailed feta assessment as soon as possible. Call 788-6240 to arrange.
8	*
9	THIS SAMPLE WAS DRAWN TOO EARLY. Please send anothe sample between 16 and 18 weeks' gestation or if this is not possible send sample at next visit.
10	THIS SAMPLE WAS DRAWN TOO EARLY. Please send another sample between 16 and 18 weeks' gestation. On the basis of the patient's age on the due date, an amniocentesis could be offered Please call 787-4804 to arrange if the patient wishes.

Table 2. (cont'd)
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Message #	Message
11	THIS SAMPLE WAS DRAWN TOO LATE (i.e., > 24 weeks) and therefore cannot be reliably interpreted. If you have concerns regarding fetal growth or anomalies, you may wish to arrange a detailed ultrasound or a fetal assessment.
12	Considering the diabetic correction, THE SAMPLE WAS DRAWN TOO EARLY. Please repeat between 18 and 20 weeks' gestation.
13	Considering the diabetic correction, THE SAMPLE WAS DRAWN TOO EARLY. Please repeat between 18 and 20 weeks' gestation On the basis of the patient's age on the due date, an amniocentesis could be offered. If desired, please call 787-4804 to arrange.
14	THE MATERNAL SERUM AFP IS LESS THAN THE MEDIAN. The risk for Down syndrome is therefore increased over the age-related risk. However, given the gestation, no accurate Down syndrome risk can be quoted. If genetic counselling and amniocentesis are desired, call 788-6240.
15	THE AFP IS CONSIDERED ELEVATED even though this is a multiple gestation pregnancy. A fetal assessment is recommended Please call 788-6240 to arrange.
16	THE AFP IS NORMAL. However, as the patient will be over age 35 by the due date an amniocentesis may be offered. Please cal 787-4804 to arrange if the patient wishes.

* Blank message, personalized message can be written later.

The protocols used by the program are reviewed regularly, and modifications are made as deemed appropriate. Previously, a repeat sample was requested before a fetal assessment was suggested for an elevated maternal serum AFP. This policy was reviewed in 1988 and discontinued because the maternal serum AFP level usually remained high, the repetition of the test increased patient anxiety, and it led to avoidable delay before fetal ultrasound assessment. In addition, one patient had a normal repeat level but had a fetus with a neural tube defect.

If a fetal assessment is recommended or an amniocentesis is offered, these appointments are made at the request of the referring physician. Counselling is provided by a member of the program team to the woman and any accompanying family members at that time. If the woman wishes, a detailed fetal assessment is done. An amniocentesis is offered for an increased Down syndrome risk. Subsequent actions depend on what is found on fetal assessment. If an abnormality is found (e.g., a neural tube

defect or fetal death), the woman is counselled in depth. If an explanation for the abnormal maternal serum AFP is found (e.g., incorrect gestational age determination, twins), the maternal serum AFP is reinterpreted. If no explanation is found for an elevated maternal serum AFP, follow-up assessments are scheduled at regular intervals throughout the pregnancy to monitor fetal growth and well-being. No follow-up is arranged for women with low maternal serum AFP levels unless there are other indications.

Review of 1990 and 1991 Statistics

Introduction and Methods

Two computer data base files were available with information derived from all samples received during 1990 and 1991. The first file contains data on information originally reported to the physician. When updated information is received, a revised report is sent to the physician. The second data base file incorporates these changes and thus represents the more accurate information. These two files were closely analyzed. All analyses refer to this two-year period. If a patient's address was not given on the requisition, the patient was assumed to live in the town in which the referring doctor practised. The frequencies of live births by health region for the years 1990 and 1991 were supplied by the Department of Vital Statistics. Statistical analyses were done using the 1990 Statistical Package for the Social Sciences (SPSS/PC 4.0).

Results and Discussion

Data files for 1990 and 1991 were analyzed, and results are shown in

the following tables.

Since the start of the Manitoba Maternal Serum AFP Screening Program in 1984, the number of women screened per year has increased steadily. The numbers have levelled off in the last two years, with approximately 10 400 women screened per year. The figure of 10 400 samples per year does not include the repeat samples handled by the program; this accounts for approximately 2 000 extra samples per year. It should be noted that 84 percent of women have only one sample taken, while 94 percent of women require two or less. Previous reviews of the program have shown a decrease in the number of women screened during the summer months; this effect was again seen.

The age distribution of referred women is shown in Table 3. Table 4 shows the number of women under 35 years of age who were given a risk for having a child with Down syndrome equal to that of a woman 35 years of age or older during the study period. Table 5 shows the number of women 35 years of age or older who were determined to be at increased risk compared to their age-related risk. In two years, 3.2 percent (676) of

women were determined to be at increased risk of having a baby with Down syndrome compared to their age-related risk, and their physicians were informed of this. Another 347 women (1.6 percent) were determined to be at increased risk, but no action was necessary as they had already had an amniocentesis or chorionic villus sampling. Of screened women under 35 years of age, 1.9 percent were given a risk of having a Down syndrome child equal to (or greater than) that of a 35-year-old. Of women under age 35, 1.8 percent were in this category in 1990 compared to 2.1 percent of women in 1991. Knight et al. (1988) stated that approximately 2.6 percent to 6 percent of women under 35 years of age should initially be given a Down syndrome risk of a 35-year-old or greater. In May 1991, we modified the protocol for estimation of Down syndrome risk. The number of women under 35 years of age who were offered an amniocentesis was closer to the "expected" frequency in 1991 than in 1990. This trend will be monitored closely in the future; however, it may be difficult to offer an amniocentesis to more women, as the cytogenetics laboratory may not be able to handle an increased workload.

Age (yrs.)	No.	%
< 25	5 277	25.1
25-29	7 678	36.5
30-34	5 616	26.7
35-39	2 152	10.2
> 39	336	1.6
Total	21 059	100.0

Risk equivalent age	Age (yrs.)								
for Down syndrome (yrs.)	27	28	29	30	31	32	33	34	Total
35.50	6	8	7	7	9	9	4	26	76
36.50				4	9	1	10	10	34
37.50				19		21	28	27	95
38.00					9				9
38.50						15	17	28	60
39.50							8	11	19
40.50								5	5
Total	6	8	7	30	27	46	67	107	298

Total Table 5. Number of Women 35 Years of Age or Older with a Risk for Down Syndrome over Their Age-S - N $\alpha - \omega$ Age (yrs.) Q 0 -4 2 2 2 3 2 3 5 7 თ -5 0 0 0 0 0 21 0 1 2 2 1 2 1 2 1 \sim S 18 22 27 27 ∞ 16 16 54 -Risk equivalent age for Down syndrome (yrs.) Related Risk 40.00 40.50 41.00 41.50 42.50 42.50 42.50 44.00 44.50 44.50 46.50 47.00 47.50 48.50 38.00 38.50 39.00 39.50 35.50 36.50 37.00 37.50 Total

The distribution of MOMs (up to 2.3) reported is shown in Figure 2; 3.2 percent of first samples were reported to have a MOM of 2.3 or greater. Most maternal serum AFP screening programs will follow between 2.5 percent and 5 percent of their population for an elevated result, indicating an increased risk of having a child with a neural tube defect (Macrae et al. 1990). The mean reported MOM value for original samples was 1.15 (standard deviation = 0.67). The median MOM was 1.0. In 2.200 cases, no MOM was calculated (i.e., sample too early, sample too late, inconsistent dates). The distribution of MOM values was as expected.

Of the patients, 5.4 percent had uncertain or irregular periods, and thus the date of the last normal menstrual period could not be used for gestational age assessment. About one-third (34.3 percent) of the women had an ultrasound before the first sample was drawn. A family history of neural tube defects or of Down syndrome was reported for 1.5 percent and 1.2 percent of the women, respectively. Table 6 shows the distribution of original samples received according to the gestation on the date the sample was drawn; 5.5 percent of original samples (1 158 samples) were drawn too early for interpretation, while 0.9 percent of samples (183 samples) were received too late. One hundred and twenty-six samples (0.6 percent of total) of the 1 158 samples received too early for interpretation were samples drawn at the time of a chorionic villus sampling procedure.

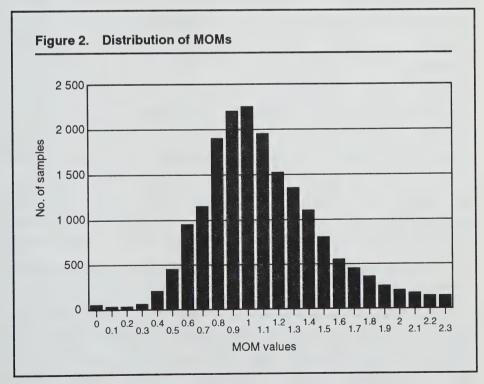


Table 6. Distribution of Gestational Ages for Original Sample						
Gestation (wks.)	No.	%				
< 12	212	1.0				

destation (wks.)	110.	
< 12	212	1.0
12-14.5	946	4.5
15-15.5	3 292	15.6
16-16.5	5 897	28.0
17-17.5	4 213	20.0
18-18.5	2 608	12.4
19-19.5	1 224	5.8
20-24	1 285	6.1
> 24	183	0.9
not determined	1 199	5.7
Total	21 059	100.0

Table 7 shows that about half of all samples came from general practitioners and somewhat fewer from obstetricians. Five hundred and sixty-seven physicians referred patients in the two-year period; only 55 physicians referred 50 or more patients per year. Of the samples received, 24.7 percent had no address; for these, the patient was assumed to live in the town where her physician practised. Table 8 shows the geographic distribution of patients. Most (70.1 percent) of the samples came from Winnipeg, while 25.5 percent came from rural Manitoba. The Manitoba program also provided testing for 793 patients referred from the University of Alberta. Table 9 compares the number of referrals to the number of births in each region in 1990 and 1991. It is clear that Winnipeg has a disproportionately high rate of referrals compared to all other regions. The percentage of women from Winnipeg may have been overestimated if many women were treated by a Winnipeg physician yet delivered outside Winnipeg, so these numbers are approximate, but Winnipeg is clearly overrepresented. The Eastman region of Manitoba is strikingly underrepresented. It is interesting to note that in our survey of Manitoba physicians, one of the most critical comments about maternal serum AFP screening came in a letter from five physicians in a practice in the Eastman region.

Rural women were more likely to have an ultrasound before maternal serum AFP screening (36.1 percent vs. 33.6 percent for Winnipeg patients, p=0.001). Non-Winnipeg physicians more often sent samples at less than 15 weeks (5.5 percent vs. 4.9 percent) or after 20 weeks (6.1 vs. 5.3 percent) (p<0.0001) of gestation. Family or general practitioners more often ordered an ultrasound first (36.7 percent) compared to obstetricians (31.8 percent) (p<0.0001). Also, they sent samples more often than obstetricians before 15 weeks (5.7 percent vs. 4.5 percent) or after 20 weeks

(6.9 percent vs. 4.2 percent) (p < 0.0001) of gestation. Physicians who referred fewer than 200 patients in the two years, compared to those who referred more, referred them more often before 15 weeks (5.7 percent vs. 4.2 percent) or after 20 weeks (6.1 percent vs. 4.5 percent) (p < 0.0001) of gestation. Physicians who referred fewer than 300 patients in the two years were also more likely than those who referred more to order an ultrasound first (36.3 percent vs. 28.7 percent) (p < 0.0001).

Table 7. Distribution of First Samples Received, by Type of Physician

Physician type	No.	%
Family/general	10 013	47.5
Obstetrician	9 815	46.6
Fetal Assessment Unit	447	2.1
Genetics	784	3.7
Total	21 059	100.0

Table 8. Frequency of First Samples Received, by Patient's Address

Address	No.	%
Alta.	. 793	3.8
BC	2	0.0
NWT	37	0.2
Ont.	54	0.3
Rural Man.	5 375	25.5
Winnipeg	14 762	70.1
Sask.	29	0.1
USA	7	0.0
Total	21 059	100.0

Table 9. Distribution of Referrals, by Manitoba Health Care Regions

Region	1990-91 referrals	Percentage of total referrals	1990-91 births	Percentage of total births	Approximate referrals/ 100 births
Central	1 058	5.2	3 074	8.9	34.4
Eastman	219	1.1	3 177	9.2	6.9
Interlake	329	1.6	1 792	5.2	18.4
Norman	350	1.7	3 089	9.0	11.3
Parklands	445	2.2	1 188	3.5	37.5
Westman	1 658	8.2	3 117	9.1	53.2
Winnipeg	16 161	79.9	18 982	55.1	85.2
Total	20 220	100.0	34 419	100.0	58.8

Review of the 1990 Patients with Abnormal Results

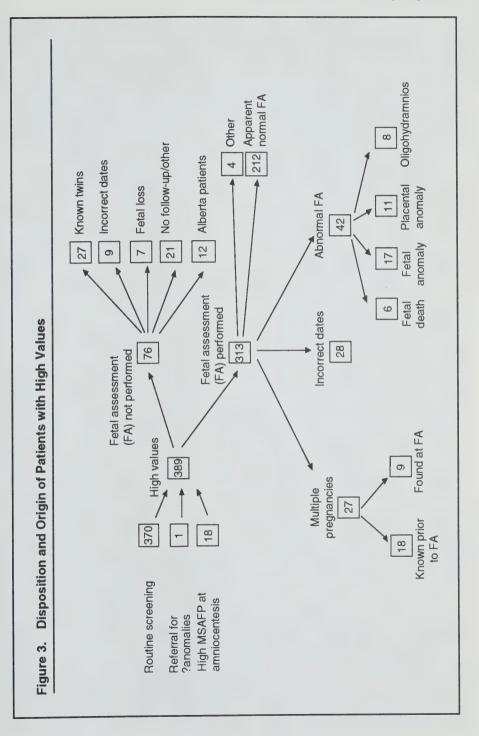
Relevant records are routinely reviewed on all patients with abnormal maternal serum AFP values. The results for patients screened in 1990 are discussed below.

In 1990, 10 362 pregnancies were screened. As shown in Figure 3, 389 (3.8 percent) had a sample that was initially considered elevated (≥ 2.3 MOM) and 441 (4.3 percent) had values that were considered low.

Patients with High Values

More than 80 percent (313/389) of the women with high values subsequently had a fetal assessment. Of the 76 women who did not have a fetal assessment, 12 were Alberta patients and were presumably followed up there. Figure 3 outlines the different consequences in these pregnancies. In the 20 women with no follow-up, either patients did not keep appointments or no referral was made by the physician.

As outlined in the lower half of Figure 3, for the women who had fetal assessment for elevated maternal serum AFP, an obvious reason was seen in 97 (31.0 percent), for example multiple pregnancy or incorrect dates. The average discrepancy for incorrect dates was an underestimate of 4.7 weeks. As shown, 42 women (13.4 percent of those having a fetal assessment) had an abnormal initial examination. An apparently normal, viable, singleton fetus was seen in 212 (67.8 percent of women undergoing fetal assessment), and in four others the fetus was difficult to see.



In all, 15 women went on to have an amniocentesis for elevated maternal serum AFP (3.7 percent of Manitoba patients with elevated levels, approximately 1 in 690 of all screened patients). These included the four whose fetus could not be adequately seen and two who were 35 years of age or older. Six other amniocenteses were performed for further evaluation of anomalies (two omphaloceles, one bilateral choroid plexus cysts, one anterior meningocele, one cystic hygroma, and one meningomyelocele), two for a placental anomaly, and one whose only indication was unexplained high maternal serum AFP. All the karyotypic findings were normal.

Table 10 shows the explanation for elevated maternal serum AFP among Manitoba patients. When women with explanations for an elevated level other than a fetal anomaly are excluded along with the patients not referred for fetal assessment, 6.0 percent of such women had a fetus with an anomaly directly detected by maternal serum AFP. These included five with meningomyelocele, five with anencephaly (including one identified in a woman who had previously declined amniocentesis for late maternal age), two with omphalocele, and one with a cystic hygroma. The women opted to have therapeutic abortions for all these fetuses except one with anencephaly where the mother continued the pregnancy and delivered a stillborn female infant at 39 weeks' gestation, one with omphalocele who died in utero at 22 weeks' gestation, and one with an omphalocele who was live born at 31 weeks' gestation and diagnosed as having Beckwith-Wiedemann syndrome. There were three other infants detected with anomalies at fetal assessment whose anomalies would not be expected to cause an elevation in the maternal serum AFP level. Two had choroid plexus cysts and in both cases healthy term infants were born. In the other case the fetal assessment revealed an incorrect gestational age assignment; however, the fetus had scoliosis and an anterior meningocele. This male was born at 40 weeks' gestation and underwent corrective surgery. Three other women among the followed patients had fetal anomalies detected at or shortly after fetal assessment. In the first, there were multiple anomalies in one twin; in the second, the gestational age was found to be underestimated but the fetus had ureteropelvic junction obstruction confirmed at delivery; and, in the third, a bladder outlet obstruction was diagnosed at autopsy in a fetus who was noted to be dead at a 15-week fetal assessment. One other woman with a fetus with anencephaly had an elevated maternal serum AFP level. However, this patient was screened late, at 28 weeks' gestation, when maternal serum AFP values are not usually reported because of less well-established normal ranges; fetal assessment revealed the anencephaly, and labour was induced.

Outcomes of pregnancy are known for 94.2 percent of patients who had a fetal assessment and for 28.1 percent of those who did not. The outcomes of the pregnancies where fetal anomalies were found have been reported above. The 10 women who had placental anomalies noted on

Table 10. Initial Explanations for High Maternal Serum AFP Values in Manitoba Patients

Elevated levels	No.	%
Multiple pregnancies	54*	14.3
Incorrect gestational age	37*	9.8
Fetal death	13*	3.4
Fetal anomaly	14**	3.7
Placental anomaly	11	2.9
Severe oligohydramnios	8	2.1
No follow-up	20	5.3
Incorrect requisition	1	0.3
No reason found	219	58.1
Total	377	100.0

* Includes one fetus with a fetal anomaly.

** Excludes three with anomalies detected on ultrasound that would not be expected to raise maternal serum AFP.

ultrasound and whose outcome was known (90.9 percent) delivered nine surviving infants. One was delivered at 35 weeks' gestation and another was growth retarded. One infant had been noted at fetal assessment to have a possible abdominal mass, and allantoic cysts were observed; after term delivery further evaluation diagnosed cross-fused renal ectopia with a dysplastic left kidney. One other pregnancy ended with a placental abruption at 23 weeks, leading to a stillbirth. Placental pathology revealed a large subchorial haematoma (Breus' mole). This was also seen in a section of the placenta from another patient in this group. The placenta on fetal assessment had been large and irregular, but the fetus developed normally and did well post-natally.

The eight patients with severe oligohydramnios detected at fetal assessment did not fare so well. Five of the women elected to terminate their pregnancy due to the poor prognosis for such cases. In the three others, premature delivery occurred at 27 to 28 weeks and the infants died in the neonatal period. In none of these cases was a renal tract disorder

identified on post-mortem examination.

Outcomes of the multiple pregnancies are known for all but one of the women seen for fetal assessment. There were 24 sets of twins and 2 sets of triplets. There were three fetal deaths (5.6 percent). Two were male triplets with a monochorionic placenta. Their female co-triplet survived despite premature delivery at 29 weeks. The other death was a twin with multiple anomalies; the co-twin did well. The mean gestational age at delivery was 35.8 ± 2.6 weeks for the multiple births as a whole, and the birthweights of the surviving infants ranged from 2 379 to 2 556 g. An

interesting finding in one case was that an ultrasound examination at eight weeks had been reported as a singleton pregnancy, and twins were detected only at fetal assessment after referral for high maternal serum AFP.

When a woman with elevated maternal serum AFP levels has no obvious reason for such levels, amniocentesis is not routinely offered if she is less than 35 years of age and the fetus is well seen. However, it is recommended that follow-up fetal assessments to evaluate fetal growth and well-being be carried out at 20, 24, 28, 32, 36, and 38 weeks' gestation in these high-risk pregnancies.

There were 196 women in this category, and although most of these pregnancies ended successfully, there was a marked increase in maternal and fetal complications in this group. Pregnancy outcomes are available for 93.9 percent and indicate a relatively high perinatal mortality rate of 5.5 percent. One other infant died of sudden infant death syndrome. Prematurity and low birthweight were more common; the average gestational age was 38.3 ± 3.4 weeks, with 17.9 percent of infants delivered before 38 weeks and 7.6 percent before 34 weeks. Mean birthweight was 3 127 ± 780 g, with 14.7 percent of infants weighing 2 500 g or less and 6.0 percent weighing 1 500 g or less. Other complications of pregnancy, such as hypertension, gestational diabetes, intrauterine growth retardation, and positive Kleihauer tests indicating maternal-fetal bleeding, were noted in several pregnancies. Placental anomalies were noted in four other women either at later fetal assessments or after delivery, and one woman was observed to have a degenerating fibroid. In three others, perinatal death was directly attributable to placental problems, with two abruptions and another rapidly growing Breus' mole. Three infants had major congenital malformations, including one with tracheoesophageal fistula and esophageal fistula (suspected at later fetal assessment due to hydramnios), one with neuroblastoma, and one with pyloric stenosis. Thus, in at least 20.8 percent of these pregnancies there was a major fetal or maternal complication affecting management. As in previous studies (Sowers et al. 1983) an excess (61 percent) of males was observed.

In some cases, such as the woman with Breus' mole, one with fetal varicella, one with chronic abruption, and one with a fall-off in fetal growth followed by an intrauterine death, early awareness of a problem did not change the outcome of the pregnancy. However, in other cases, the protocol for follow-up of these women in later pregnancy may have led to an improved outcome. For example, during one routine 38-week scan, oligohydramnios was found and labour was induced. During another, at 32 weeks, the woman was noted to be hypertensive with a growth-retarded fetus and was admitted to hospital for management. Labour was induced in three other women whose fetuses had shown significant fall-off in growth between their appointments at 34 and 38 weeks, and one woman with systemic lupus erythematosus whose fetus had continued to grow (albeit along the 10th percentile) was delivered at 30 weeks' gestation of a 800 g

infant. All of these infants did well and had minimal neonatal complications.

Outcomes are also available for 14 of the 18 women who had amniocentesis for unrelated reasons but who were found to have an elevated maternal serum AFP when the blood sample was taken before amniocentesis. In one case, twins were determined on fetal assessment before amniocentesis, and in one other an additional empty gestational sac seen during an ultrasound at eight weeks could have led to high maternal serum AFP levels. Pregnancy outcomes are not yet available for these cases. Data on the 14 singleton pregnancies with known outcomes indicate that the experiences of these women are similar to those first identified by routine maternal serum AFP screening. Four fetuses (28.5 percent) had intrauterine growth retardation and three had minor anomalies. One other infant had a 45,X/46,XX karyotype. One mother developed hypertension and one had a positive Kleihauer test. All of these infants did well. However, 1 of the 14 women had a fetal loss. This woman had two borderline (2.1 MOM) maternal serum AFP levels before deferred amniocentesis for advanced maternal age was done. Fetal assessment at that time was normal and so was the fetal karyotype. However, very elevated (4.7 MOM) maternal serum AFP and amniotic fluid AFP levels led to further fetal assessments over the next few weeks. These showed a rapidly growing placental haematoma and the pregnancy ended with an intrauterine death at 23 weeks' gestation. Again, Breus' mole was detected on placental pathological examination. The fetal assessment done before amniocentesis was especially detailed in this case, as the mother had reported at her prenatal counselling session that she had used cocaine in early pregnancy. Thus, these women seem, like any others with elevated maternal serum AFP, to be at risk for pregnancy complications. The mean gestational age at delivery was 39.4 ± 1.5 weeks in the 13 surviving infants, and their birthweight was 3 220 ± 67 g; six were males, seven females.

As shown in Figure 3, apart from the 212 women (196 from routine screening and 16 from maternal serum AFP screening at the time of amniocentesis done for advanced maternal age) whose initial fetal assessments did not indicate an obvious cause for abnormal maternal serum AFP, there were four others for whom the fetus had not been seen adequately to rule out anomalies. Amniocentesis in all cases revealed normal karyotypes and biochemical parameters. One woman who developed gestational diabetes and hypertension delivered a 2 050 g female at 33 weeks, one fetus demonstrated intrauterine growth retardation on later fetal assessments and weighed 2 420 g on delivery at 39 weeks, and the third woman developed gestational diabetes but the fetus did well. The fourth had three positive Kleihauer tests indicating possible placental haemorrhage, and a male fetus weighing 3 495 g was born at 38 weeks' gestation with mild foot deformities. Thus, the frequency of pregnancy complications in this group was similar to that in the larger group of 212 women.

Pregnancy outcomes are not yet available on women with normal maternal serum AFP values from 1990; when they are received they will of necessity be less detailed than the reviewed chart patients. Thus, it is not vet possible to determine directly if the frequency of complications in women with high maternal serum AFP in 1990 was significantly greater than in those with normal values screened in 1990. However, the preliminary data and our experience from data analyses from previous years and from literature reviews (Burton 1988; Katz et al. 1990; Read et al. 1980; Simpson et al. 1991) would indicate this to be so. The small group of 29 women with incorrect gestational age estimations and known pregnancy outcomes can provide a useful comparison, as they had a similar detailed chart review (Table 11). Even this comparison group may have a higher than average frequency of pregnancy complications, as they still have higher than average maternal serum AFP levels even after correction for gestational age (mean 1.5 ± 0.4 MOM), and the fact that their dates were inaccurate may indicate a younger, more vulnerable population. However, when comparing the two groups, it is apparent that the women with unexplained high maternal serum AFP tend to have smaller infants at earlier gestations than those with incorrect gestational ages. Also, the perinatal death rates are higher and oligohydramnios and hypertension were slightly higher in the high maternal serum AFP group, though these differences, like those of weight and gestation, were not significantly different. Major complications were noted in two pregnancies in the incorrect dates group (6.7 percent) compared to 20.8 percent in the women with high maternal serum AFP. The sex ratio, though it included more males, was not significantly different from expected in the incorrect dates group.

The final group of women that should be commented on are those for whom a fetal assessment was recommended but who either declined further investigation or were not referred for further testing by their physicians. So far, we have outcomes in 9 (45 percent) of the 20 women with no prenatal follow-up. Again, the figures for complications are similar to those seen in the followed groups. One woman delivered monozygous twins at home and both died. Another delivered a 651 g fetus at 23 weeks that died in the neonatal period. A third growth-retarded baby had an imperforate anus. One woman developed gestational diabetes and hypertension; her large-for-gestational-age fetus was delivered at 38 weeks' gestation and had a pneumothorax and hypoglycaemia. Mean gestational age for the seven surviving infants was 39.1 ± 1.3 weeks and mean birthweight 3.194 ± 563 g.

Thus, even in women with high maternal serum AFP where a neural tube defect is ruled out, the possibility of fetal and maternal complications appears increased. Although the potential benefit of follow-up of women with unexplained levels of maternal serum AFP remains controversial (Cunningham and Gilstrap 1991; Simpson et al. 1991), reassurance of a likely normal outcome in such cases may be premature. The Manitoba

Table 11. Outcomes of Pregnancy in Routinely Screened Women with Unexplained High Maternal Serum AFP (i.e., Normal First Fetal Assessment) and Those with Overestimated Gestational Age and Singleton Pregnancies

	Unexplained high maternal serum AFP	Incorrect dates	
	N = 184	N = 29	
Mean birthweight (g)	3 127 ± 770	3 323 ± 1.8	
≤ 2 500 g (%) ≤ 1 500 g (%)	14.7 6.0	3.4 0.0	
Mean gestational age (wks.)	38.3 ± 3.4	39.3 ± 1.8	
≤ 37 weeks (%) ≤ 33 weeks (%)	17.9 7.6	10.3 0.0	
Intrauterine growth retardation (%)	10.3	3.4	
Perinatal deaths (%)	5.5	0.0	
Oligohydramnios (%)	3.8	0.0	
Hypertension (%)	5.5	3.4	
Gestational diabetes (%)	3.3	6.9	
Infant anomalies major minor	1.6 8.7	3.4 6.8	
Sex ratio infants (M:F)	1.58	1.63	
Major maternal/fetal complications (%)	20.8	6.7	

maternal serum AFP program, because of its ability to follow up and document outcomes in these women, may be a unique resource for evaluating the answer to this question. Currently, we are analyzing maternal serum AFP values at later gestations (i.e., 25 to 40 weeks) in women who had normal maternal serum AFP values on initial screening. When such ranges of normal values are available, we will be able to re-evaluate changing patterns of maternal serum AFP in women with initially high maternal serum AFP to determine the types of patterns, if any, that indicate which women might benefit from further follow-up and which can be reassured and have follow-up discontinued after the scan at 20 or 24 weeks.

At our cut-off of 2.3 MOM in Manitoba in 1990, a woman with a high maternal serum AFP level who had not already been determined as having a twin pregnancy had about a 1 in 2 chance of having an essentially uncomplicated pregnancy and delivering a normal newborn, a 1 in 9 chance of having overestimated dates, a 1 in 16 chance of a major fetal abnormality, a 1 in 24 chance of an early fetal loss, a 1 in 26 chance of a perinatal death, a 1 in 35 chance of an unsuspected multiple pregnancy, and a 1 in 39 chance of severe oligohydramnios. For women whose initial maternal serum AFP levels are 4.0 MOM or greater, the prognosis is even more serious. None of these pregnancies was without complication; only 10 of 35 women had a surviving infant and 4 (11.4 percent) had fetuses with neural tube defects.

Patients with Low Values

Our protocol in 1990 considered patients with low values in one of two categories: those with a maternal serum AFP value of \leq 0.4 MOM, and those with a value of \leq 0.7 MOM whose age combined with their maternal serum AFP value gave them an increased risk of having a live-born child with Down syndrome of greater than or equal to 1 in 200. A total of 441 women fell into one or both of these categories. All women at increased risk for Down syndrome were offered amniocentesis (or cordocentesis in more advanced gestations) unless a prenatal diagnosis had been made previously. For women with low values but not at significantly increased risk of Down syndrome, a fetal assessment was recommended if the maternal serum AFP level was \leq 0.25 MOM, while a repeat sample was requested if levels were between 0.25 and 0.45 MOM. If the maternal serum AFP level remained low, a dating scan was recommended, but if this confirmed or corrected the gestational age, no further follow-up was implemented.

Figure 4 shows the disposition of the 105 women with values ≤ 0.4 MOM who were not at increased risk for Down syndrome (1.0 percent of all screened pregnancies). By definition all of these women were under 30 years of age. Most (68.5 percent) of these women were not followed by the maternal serum AFP program, probably because a reason for the low maternal serum AFP was found and a repeat sample was not sent. In 20.2 percent of the cases with values at 0.25 to 0.4 MOM, a repeat sample was normal (i.e., > 0.4 MOM). This was often due to testing before or close to the recommended earliest gestation of 15 weeks for maternal serum AFP screening, when the normal rise in maternal serum AFP is becoming apparent. When Manitoba patients with unexplained very low levels of maternal serum AFP (i.e., ≤ 0.25 MOM) were seen for fetal assessment (75 percent), a reason for the very low levels was identified in all: three had molar pregnancies, two had missed abortions, and four had overestimated gestational ages. Table 12 gives the initial evaluation of reasons for low maternal serum AFP in these young women. Overestimated gestational

age was a common cause (18.3 percent); this rose to 29.2 percent in patients where we have some follow-up. The average overestimation in these cases was 4.5 weeks. False pregnancy, molar pregnancy, and impending fetal loss accounted for 5.8 percent (9.2 percent of those with some follow-up) and were confined to women with very low values.

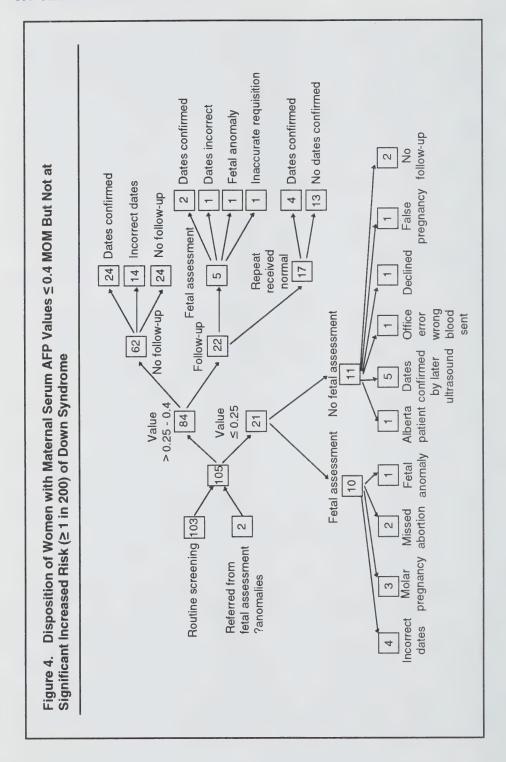
The two women who had low maternal serum AFP values determined after fetal assessment detected anomalies had amniocentesis. One had a fetus with Down syndrome, the other a fetus with trisomy 18. In both cases the samples were taken at gestations over 30 weeks; they had liveborn children later.

Outcomes of pregnancy are known for a relatively small number of the other young women with low values and viable pregnancies because few had fetal assessments and we do not have data on their outcomes. All have had live-born children without serious complications except for one insulin-dependent diabetic woman with incorrect dates who had a 905 g infant at 26 weeks' gestation, who did well.

Table 12. Initial Evaluation of Manitoba Women with Maternal Serum AFP Values \leq 0.4 MOM But Not at Significant Increased Risk (\geq 1 in 200) of Down Syndrome

Low levels	No.	%
Fetal anomalies*	2	1.9
Incorrect dates	19	18.3
Molar pregnancy	3	2.9
Missed abortion	2	1.9
False pregnancy	1	1.0
Incorrect requisition/office error	3	2.9
Unexplained confirmed low values	35	33.7
No follow-up	39	37.5
Total	104	100.0

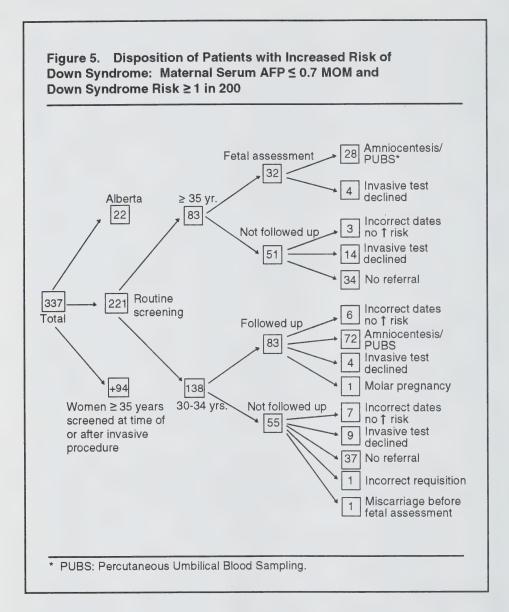
^{*} Patients referred from Fetal Assessment Unit.



There were 337 women (3.3 percent of the screened population) whose maternal serum AFP value in conjunction with their age gave them a risk for having a live-born child with Down syndrome greater than that of a 36-vear-old. Ninety-four of these women had already had a chorionic villus sampling test or had a maternal serum AFP sample taken at the time of amniocentesis for late maternal age. An additional 22 women were from Alberta and presumably followed up there. Thus, there were 221 routinely screened Manitoba patients in this group. The disposition of these patients can be seen in Figure 5. In all, 138 (62.4 percent) were between 30 and 34 years of age and thus would not normally have been offered prenatal diagnosis in the screened pregnancy. Table 13 summarizes the results of initial evaluations in these women. Among those 35 years of age or older, 33.7 percent had an invasive test performed while 21.7 percent are known to have declined further testing. At least 5 of those who had testing had declined testing previously but changed their minds when the maternal serum AFP results were available. In 41 percent of cases no referral for amniocentesis was made. We do not know if these women declined to have further evaluation or if they were not informed of the results of maternal serum AFP screening. Among the 138 women under 35 years of age, a higher proportion was referred for further testing and 52.2 percent subsequently had an invasive test. Some women were found to have incorrect gestational age estimations at fetal assessment, and corrected maternal serum AFP interpretations no longer placed them at increased risk. Such reassignments of gestational age were made conservatively in this group due to the possibility of considering a pregnancy with a growth-retarded Down syndrome fetus to be less further along than expected.

Table 13. Evaluation of Women with Increased Risk of Down Syndrome: Maternal Serum AFP \leq 0.7 and Down Syndrome Risk \geq 1 in 200 Ascertained by Routine Screening

	No.	%
Patients 35 years of age or older (N = 83)		
Invasive test performed	28	33.7
Invasive test declined	18	21.7
Incorrect dates, risk not increased by maternal serum AFP	3	3.6
No referral for invasive testing	34	41.0
Patients under 35 years of age (N = 138)		
Invasive test performed	72	52.2
Invasive test declined	13	9.4
Incorrect dates, risk not increased by maternal serum AFP	13	9.4
No referral for invasive testing	37	26.8
Other (molar pregnancy, fetal loss, incorrect requisition)	3	2.2



Outcomes for these women can be considered in two ways. First, one can examine the frequency of chromosomal anomalies in these groups. Of the women undergoing invasive testing, 3 of the 28 over 34 years of age had fetuses with chromosome anomalies (one trisomy 18, one trisomy 21, and one inversion of chromosome 6). Among the 72 women 30 to 34 years of age, 1 had a fetus with trisomy 21 (1.3 percent or 1 in 72). Among the 94 women with low maternal serum AFP who had already opted for

prenatal diagnosis, 2 had a fetus with Down syndrome and 2 had other chromosomal anomalies detected: a deletion of 14p and a reciprocal translocation. We do not yet know the outcome of all pregnancies for the women whose charts were not available because they did not have fetal assessment, but we know of two other cases of aneuploidy. One late maternal age patient was not referred for further testing and had a trisomy 18 fetus at 34 weeks' gestation. Another who had declined testing had a fetus with multiple anomalies due to trisomy 18, which had been discovered on ultrasound at 31 weeks; the dates were incorrect, and the maternal serum AFP had been readjusted to 0.9 MOM. This was the fetus with a small neural tube defect discussed later in this report. Of the five fetuses detected prenatally with aneuploidy, the mothers elected to terminate the pregnancy in three of the four trisomy 21 cases. In the fourth trisomy 21 case the pregnancy continued to 37 weeks, when a liveborn infant with a congenital heart defect was delivered. The infant with trisomy 18 was live-born at 42 weeks' gestation and is still living.

Trisomy 21 or 18 was therefore determined in the fetuses of five women who had prenatal testing due to increased risks based on their maternal serum AFP levels. This figure of 1 in 39 is higher than the 1 in 81 in the total population in Manitoba screened by chorionic villus sampling or amniocentesis in 1990. The frequency of aneuploidy in women whose maternal serum AFP levels were not equal to or below 0.7 MOM was 1 in 134. Among the late maternal age tests in Manitoba in 1990, the frequency of trisomy 18 or 21 detected at amniocentesis was identical to the 1 in 72 seen in women under 35 years of age tested because of low maternal serum AFP. We will be able to generate more specific figures concerning the sensitivity and specificity of maternal serum AFP screening for aneuploidy when information is available on pregnancy outcomes in the women who declined testing or were not referred for further investigations.

Second, with respect to other complications of pregnancy, the moderate reductions of maternal serum AFP observed in most of these patients do not seem to compromise the mother or fetus. We have outcomes on 48.2 percent of patients who were followed up and no losses have been reported after amniocentesis. The infants with normal karyotypes have all been live-born, with an average birthweight of $3\,431\pm463\,g$ and a gestational age of 39.7 ± 3.4 weeks. These values are significantly (p < 0.01) higher than those in the infants followed up because of high maternal serum AFP. The sex ratio showed a non-significant excess of females: 24 males to 30 females. Only one child had another major congenital anomaly: craniosynostosis.

In 1990, as in previous years, we observed the association of low maternal serum AFP levels with fetal loss and incorrect gestational age noted by other researchers (Bennett et al. 1979; Burton 1988; Davenport and Macri 1983; Haddow et al. 1987; Kjessler et al. 1977a; Simpson et al. 1987). Previously, we had noted an association between very low (≤ 0.25 MOM) maternal serum AFP and large-for-gestational-

age infants (Evans et al. 1990b), but we do not yet have birthweight information for the 1990 cohort with continuing pregnancies and confirmed low maternal serum AFP values to see if the association persisted in this population.

Our frequency of 1 in 72 for aneuploidy in younger women with increased risks made on the basis of their age and maternal serum AFP levels is also similar to that observed by others. For example, DiMaio et al. (1987), using a cut-off of risk equivalent to 35 years, found a rate of chromosomal anomalies of 1 in 112, while Baumgarten et al. (1985), using a similar cut-off, found frequencies of 1 in 74 for Down syndrome and 1 in 63 for all trisomies. Other workers have also observed that other chromosomal anomalies may be more common in women referred for low maternal serum AFP values (Ben-Yishay et al. 1988; Drugan et al. 1989; Rao and Atkin 1988; Redwine et al. 1988). This was seen in 1990 among our late maternal age patients with low maternal serum AFP but not in the small number tested who were under 35 years of age; we have also noted this phenomenon in previous years, especially with respect to Turner syndrome mosaics (Evans et al. 1990a).

In summary, women with low maternal serum AFP levels are at increased risk for fetal loss and overestimated gestational age. Such women may also be at increased risk to have aneuploid fetuses, and maternal serum AFP can be used with maternal age to improve the sensitivity of prenatal diagnostic screening for Down syndrome from about 20 percent to 35 percent (L. Anders and J.A. Evans, unpub. data 1992; Cuckle et al. 1987; Lippman and Evans 1987; New England Regional Genetics Group 1989). Introduction of other biochemical markers, including beta-hCG and conjugated estriol (Bogart et al. 1987; Canick et al. 1988; Wald et al. 1988a), can potentially improve the sensitivity of serum screening tests still further. Meanwhile, in contrast to women at increased risk based on high maternal serum AFP, these women can be offered amniocentesis and be reassured that, if the fetal karvotype is normal, the relatively reduced maternal serum AFP values of 0.25 to 0.7 MOM do not seem to increase risks for other pregnancy complications, although birthweights may be greater.

Review of the Rates of Neural Tube Defects in Manitoba

Introduction and Methods

Manitoba is an ethnically diverse province with a high proportion of individuals of British and continental European descent. The frequency of neural tube defects is known to be high in Britain, especially in Ireland, Wales, and Scotland (corrected prevalence rates 2.4 to 3.8 in 1 000 births and terminations), but considerably lower in continental Europe (1.2 in 1 000) (EUROCAT Working Group 1991). In both Britain and continental

Europe, the birth prevalence of neural tube defects has declined in recent years. In Britain, this has been due partly to prenatal diagnosis of neural tube defect and the termination of these pregnancies, but, importantly, the total prevalence, including live births, stillbirths, and pregnancy terminations, has also shown significant secular decline since the 1960s (Cuckle et al. 1989; Davis and Young 1991; Laurence 1985; Stone et al. 1988). However, in continental Europe the total prevalence has remained stable. As part of ongoing evaluation of the Manitoba Maternal Serum AFP Screening Program, the prevalence of neural tube defects in the province since 1979 was assessed to determine both the total and the birth prevalence of these disorders and whether these parameters have changed over time.

Data from several sources, including the Manitoba Congenital Anomalies Registry, the Section of Clinical Genetics, Health Sciences Centre, and the Maternal Serum AFP Screening Program, were used to ascertain all fetuses and infants identified as having anencephaly or spina bifida since 1979. Cases of iniencephaly were included, but cases of encephalocele were not. Cases were included regardless of whether the neural tube defect was isolated or associated with other anomalies or part of a syndrome.

For each case it was determined whether anencephaly or spina bifida had been identified at birth or prenatally and, if prenatally diagnosed, by which method (i.e., maternal serum screening, amniocentesis, or ultrasound). For infants or fetuses delivered from 1982 onwards, it was determined if the pregnancy had been screened by maternal serum AFP determination or not. If prenatally diagnosed, the outcome of the pregnancy was determined (i.e., live birth, stillbirth, induced or spontaneous abortion).

Both the birth and the adjusted (i.e., births plus abortions) prevalence rates of neural tube defects were calculated for each year from 1979 to 1990 inclusive, for the two 6-year periods 1979 to 1984 and 1985 to 1990, and for the total 12-year period. For each year since 1982 the number of pregnancies screened in Manitoba by maternal serum AFP was determined and the number of neural tube defects detected and missed by screening ascertained. These figures were compared with the numbers expected among the screened patients in each year. Results are discussed below.

Results and Discussion

Table 14 shows the birth and adjusted prevalence rates by years. An average of 22 cases (births plus terminations) per year was observed over the period 1979 to 1990, and no obvious decline in total prevalence was documented. However, as can be seen from this table, and as is graphically illustrated in Figure 6, the birth prevalence started to decline rapidly relative to total prevalence in 1987.

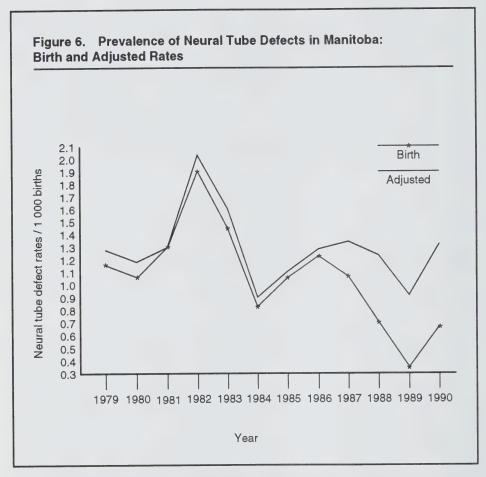


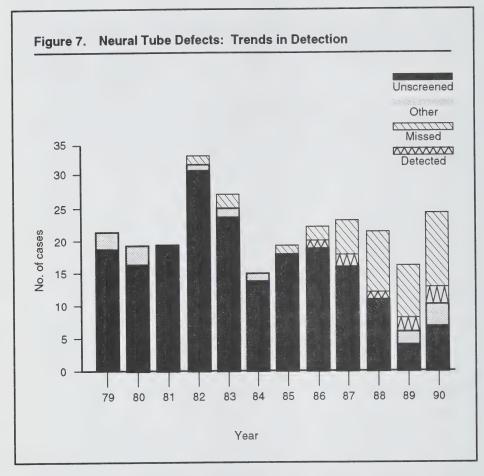
Figure 7 shows the proportion of cases detected by maternal serum AFP screening, those screened but missed, those prenatally diagnosed by other methods, and those unscreened and ascertained at birth.

Before 1985 almost all infants with neural tube defects were live-born or stillborn after 20 weeks' gestation. Occasionally, a fetus with such anomalies was detected earlier at amniocentesis or ultrasonographic examination because of a family history of neural tube defects (five cases) or for unrelated reasons, usually advanced maternal age (two cases). In 1982, the pilot program for maternal serum AFP screening was initiated in Manitoba (Evans et al. 1986). During the pilot program, which continued until October 1984, 2 045 pregnancies were screened and three fetuses with anomalies were detected. These cases represented 2.3 percent of neural tube defect fetuses identified pre- or post-natally from 1982 to 1984 inclusive. Since the introduction of a provincial program in 1985, the number of screened women has increased from 2 941 in 1985 to 10 362 in

Table 14. Birth and "Adjusted" Prevalence Rates of Neural Tube Defects in Manitoba, 1979 to 1990

Birth Total births		Total bir	th prevalence	"Adjusted" prevalence (births plus terminations)		
years	(live & still)	No.	Rate/1 000	No.	Rate/1 000	
1979	16 390	19	1.16	21	1.28	
1980	16 127	17	1.05	19	1.18	
1981	16 222	21	1.29	21	1.29	
1982	16 223	31	1.91	33	2.03	
1983	16 723	24	1.44	27	1.61	
1984	16 780	14.	0.83	15	0.90	
1985	17 220	18	1.05	19	1.10	
1986	17 112	21	1.23	22	1.29	
1987	17 064	18	1.05	23	1.35	
1988	17 129	12	0.70	21	1.23	
1989	17 445	6	0.34	16	0.92	
1990	18 011	12	0.67	24	1.33	
Total	202 446	213	1.05	261	1.29	

1990. The increased number of screened pregnancies has meant that a larger proportion of fetuses with neural tube defects has been detected prenatally. In 1985, 5 percent of pregnancies where the fetuses had neural tube defects were screened. This figure rose to 14 percent in 1986, 48 percent in 1988, 63 percent in 1989, and 58 percent in 1990. Interestingly, two of the babies born in 1989, one born in 1990, and another born in 1991 were from the same small First-Nation band in rural Manitoba. Although this unusual cluster of cases has been investigated in detail with family interviews and parental karyotyping, no genetic or environmental causes for this pocket of high frequency have been identified. None of the four cases had been screened prenatally.



In total, 48 pregnancies with a fetus with neural tube defects have been screened since maternal serum AFP was introduced as a screening test in Manitoba. The sensitivity of the test overall has been 95.2 percent for anencephaly and 79.2 percent for spina bifida. In one case of meningomyelocele, the initial maternal serum AFP level was elevated, but the repeat sample was below the cut-off for further investigation. This unusual case in 1988 led to a change in protocol whereby it was recommended that all women with a high first sample receive counselling and detailed fetal assessment. This improved the sensitivity of the test for spina bifida from 75 percent for the period 1985-88 to 83 percent for the period 1989-90. In all, nine cases of neural tube defects have occurred in screened pregnancies but were "missed." The single case of anencephaly and four cases of spina bifida, including the one mentioned above, were below the cut-off for further action. One other case was screened at 7 weeks and, despite the standard recommendation for a repeat sample to be sent at 16 to 18 weeks' gestation, none was received. Two infants had

closed defects and thus levels of maternal serum AFP below the cut-off. One infant had a small meningomyelocele and other anomalies associated with trisomy 18. This infant was born to a woman who had declined amniocentesis for late maternal age. Her maternal serum AFP level had been considered low initially and she had a fetal assessment done at 14 weeks. The ultrasonographic examination was considered normal at this time and her maternal serum AFP value was re-interpreted to be 0.9 MOM, as incorrect information had been provided on gestational age. Her maternal serum AFP age-adjusted risk for aneuploidy was 1 in 75. At 32 weeks, repeat fetal assessment revealed multiple anomalies and an amniocentesis showed trisomy 18. A repeat maternal serum AFP at this time was not elevated, and acetylcholinesterase activity was minimally elevated with a ++ positive band. This case illustrates the potential difficulty in interpreting maternal serum AFP levels in rare cases where conflicting factors serve to raise (spina bifida) and lower (aneuploidy) maternal serum AFP in the same patient.

All 19 women who were found to be carrying a fetus with spina bifida through maternal serum AFP screening sought a termination of the pregnancy. Fourteen of the 20 women with anencephalic fetuses also sought therapeutic abortions, and one other had a spontaneous loss. The five others continued their pregnancies. Two women were considered too far advanced in their pregnancies for termination due to being screened later than 24 weeks; these infants died in the neonatal period. In two other pregnancies there was a normal co-twin and the pregnancies ended near term. One of the anencephalic infants died at three days of age and the other was stillborn. In the last case the mother declined a termination of pregnancy and had a stillborn infant near term.

Most women who were found to be carrying affected fetuses chose to seek termination of their pregnancies; this has meant that the number of infants born after 20 weeks' gestation in Manitoba with neural tube defects has declined since 1985. In 1985, 95 percent of infants ascertained as having an open neural tube defect were born usually at or near term; by 1990, this figure had dropped to 38 percent.

As screening became more widespread in Manitoba, the proportion of fetuses with neural tube defects detected early in gestation was expected to increase due partly to each pregnant woman having a greater chance of being screened. Also, it was thought that more widely used screening would include less affluent women who may be at greater risk for neural tube defects. This, in fact, has occurred, as the ratio of observed numbers of neural tube defects to be expected among screened patients has tended to increase with time (Table 15).

Table 15. Number of Screened Pregnancies and Observed and Expected Numbers of Neural Tube Defects Detected in Manitoba, 1979 to 1990

	No. of	No. of neural tube defects				
	screened pregnancies	Detected	Missed	Observed total	Expected	% Observed/ expected
1982	878	1	0	1	1.78	56.2
1983	1 328	2	0	2	2.13	93.9
1984	1 168	0	0	0	1.05	0.0
1985	2 941	1	0	1	3.24	30.9
1986	4 798	2	1	3	6.19	48.5
1987	6 716	5	2	7	9.07	77.2
1988	8 450	9	1	10	10.39	96.2
1989	9 231	8	2	10	8.49	117.8
1990	10 362	11	3	14	13.78	101.6

Physician Survey

Introduction and Methods

Physicians who provide primary obstetric care play an important role in the Manitoba Maternal Serum AFP Screening Program. These physicians provide patients with their first contact with maternal serum AFP screening. They are also the ones who must first explain abnormal results to the patients. Assessment of the views and knowledge of these physicians is therefore crucial in our understanding of the program as a whole, and in understanding what needs to be done to ensure it meets the needs of the women and families it serves.

A questionnaire was developed that would ask questions about (1) demographics; (2) physicians' knowledge regarding prenatal diagnosis, including maternal serum AFP; (3) physicians' practice with respect to prenatal diagnosis and maternal serum AFP testing; and (4) physicians' views and opinions on this topic. The first question asked what percentage of the physician's practice related to obstetrics (where he/she would see pregnant women); if the answer was zero, the physician was asked to return the questionnaire with the remaining questions unanswered. However, such physicians could provide any written comment that they wished. This procedure allowed a response rate to be calculated and also enabled the program to concentrate on physicians who provide obstetric care. The survey is attached in Appendix 4.

Mailing labels were obtained from the College of Physicians and Surgeons of Manitoba for all physicians classified as obstetricians/ gynaecologists (59) or general/family practitioners (887). Approval for the survey was obtained from the University of Manitoba Ethics Committee. On 29 January 1992, questionnaires were sent to these physicians with a covering letter. Questionnaires were numerically coded to keep track of non-respondents but not to identify respondents. Three weeks after the initial mailing, a follow-up mailing was sent to all non-respondents. One week later an attempt was made to determine if the persistent nonrespondents included obstetrics in their practice. The records of the Manitoba Maternal Serum AFP Screening Program were reviewed. If the physician had sent at least one sample for maternal serum AFP testing in the past, it was assumed that he or she practised obstetrics. If no sample had been received, telephone directories were checked for the office number of that physician. If no office phone number was found, the physician was assumed not to practise obstetrics; if a phone number was available, the office was contacted to see if the doctor did practise obstetrics.

Two questions were asked about physicians' knowledge of prenatal diagnosis. For analysis of knowledge versus other factors, one point was given for each correct answer (e.g., the age at which an amniocentesis is offered is 35 years, and age is based on the due date). Similarly, three questions were asked regarding physicians' knowledge of maternal serum AFP testing. One point was given if the physicians stated maternal serum AFP is 70 percent to 90 percent sensitive for spina bifida and one point if they said maternal serum AFP was 20 percent to 40 percent sensitive for Down syndrome. One point was given for each other condition detectable by maternal serum AFP testing up to a maximum of three.

Three comments were given with a frequency of greater than 20 percent: (1) patients decline AMA (advanced maternal age) counselling because they wouldn't abort; (2) maternal serum AFP screening creates anxiety in patients and/or physicians; and (3) there are too many false-positive results with maternal serum AFP screening. As outlined in the results, comparisons were made between the distribution of these comments and various physician characteristics and responses. Chisquared tests, *t*-tests, or ANOVA were used as appropriate to check for statistical significance. For the purpose of this analysis, statistical significance was defined as a p value less than 0.01. Only significant associations will be reported. Data analysis was done using the 1990 Statistical Package for the Social Services.

Results and Discussion

A total of 642 responses was received. Excluding the nine physicians who were unavailable or whose address was incorrect, the overall response rate was 68.5 percent. One physician was phoned and refused to comment • whether or not obstetrics was part of his or her practice.

Five respondents who did not practise obstetrics provided comments (Appendix 5). All subsequent results refer to the 289 respondents who did practise obstetrics. The characteristics of the physicians who do some obstetrics are shown in Tables 16 and 17.

Table 16. Characteristics of Physicians Who Practise Obstetrics

	No.	%
Type of physician		
Family or general practitioner	251	86.9
Obstetrician/gynaecologist	33	11.4
Subspecialist obstetrician/gynaecologist	5*	1.7
Total	289	100.0
Sex		
Not given	12	4.2
Female	91	31.5
Male	186	64.4
Total	289	100.0
Practice location		
Not given	3	1.0
Winnipeg	131	45.3
Within 60 miles of Winnipeg	32	11.1
Beyond 60 miles of Winnipeg	123	42.6
Total	289	100.0

^{* 3} perinatologists, 1 paediatric adolescent specialist, and 1 reproduction specialist.

Ages ranged from 25 to 72 years, with an average of 40.8 years; years in practice ranged from 2 to 47, with an average of 15.2 years.

The response rate for this survey is comparable to that of other surveys about physician attitudes to prenatal diagnosis (Fahy and Lippman 1988; Firth and Lindenbaum 1992). Although any survey with a response rate of less than 100 percent cannot be guaranteed to be representative of the targeted population, we are not aware of any specific response bias. The views expressed seem to represent the range of opinions held by physicians.

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Religion	No.	%
Not given	71	24.6
Roman Catholic	45	15.6
Protestant	32	11.1
Anglican	19	6.6
Vone	19	6.6
Christian	18	6.2
United Church	16	5.5
Jewish	14	4.8
Mennonite	12	4.2
Hindu	9	3.1
Moslem	6	2.1
Dutch Reform	5	1.7
3aptist Saptist	4	1.4
_utheran	4	1.4
Agnostic	3	1.0
Bahai	3	1.0
Atheist	2 2	0.7
Buddhist	2	0.7
Evangelist Protestant	1	0.3
Greek Orthodox	1	0.3
Mormon	1	0.3
Orthodox	1	0.3
Pentecostal	1	0.3
otal	289	100.0

The answers to the questions testing physician knowledge regarding prenatal diagnosis or maternal serum AFP are shown in Tables 18 through 22. One of the most important findings of this survey is the clear need for more physician education about prenatal diagnosis in general and maternal serum AFP testing specifically. Disturbingly, over 16 percent of respondents did not know that a woman is considered a candidate for prenatal diagnosis if she will be 35 years of age. Only 34.9 percent knew that this age is calculated according to the woman's estimated due date. Several physicians commented that the difference between calculating the age based on the last period or on the due date was a trivial issue; thus, many women who are actually candidates for prenatal testing may not be referred because the physician is not aware of this "trivial issue." Physicians were generally aware of the sensitivity of maternal serum AFP screening for spina bifida, which is estimated to be 70 percent to 85 percent for open lesions (Macrae et al. 1990). On average, however, physicians overestimated the sensitivity of maternal serum AFP screening for Down syndrome. If an

amniocentesis was done on the 5 percent of women at highest risk on the basis of their age and maternal serum AFP level, the detection rate would be 35 percent (Wald et al. 1988b). This is lower than the estimated 50 percent sensitivity according to respondents of this survey. To achieve a detection rate of 50 percent on the basis of age and maternal serum AFP, an amniocentesis would have to be done on 12 percent of the population (ibid.), a rate much higher than the current practice in Manitoba.

Table 23 shows most physicians refer all eligible women for prenatal diagnosis but, disturbingly, some never refer, and one in seven (14.9 percent) refers or offers referral only to some eligible women. The mean estimated percentage of patients who declined referral for AMA counselling was 31.1 percent (± 28.2 percent) (range 0 percent to 100 percent). Table 24 shows that 37.7 percent of physicians offer maternal serum AFP to all patients and it is done if consent is given; 19.4 percent do it automatically unless the woman specifically declines. It is disturbing that some physicians do not offer it, some just offer it to specific patients, and in more than one in five cases, the patient has no opportunity to decide if she would like to have the test, as it is done without specific consent. The type of information supplied to the patient before maternal serum AFP testing is shown in Table 25, with the majority given verbal information only, even though information pamphlets are available from the program. Two hundred and thirty-three physicians who discuss maternal serum AFP testing with their patients estimated the time spent on this issue: the mean estimate was 4.9 minutes (± 3.4 minutes); the minimum amount was 0.5 minutes and the maximum was 22.5 minutes.

Table 18. Age at Which Physicians Stated Women Are Eligible for Amniocentesis or Chorionic Villus Sampling

Age	No.	%
20.0	1	0.3
30.0	1	0.3
32.0	1	0.3
33.0	1	0.3
34.5	1	0.3
35.0*	235	81.3
36.0	16	5.5
36.5	1	0.3
37.0	9	3.1
38.0	2	0.7
40.0	13	4.5
No response	8	2.8
Total	289	100.0

^{*} Correct response.

Table 19. Knowledge of Physicians Regarding Landmark for Age Eligibility for Prenatal Diagnosis

"Age of eligibility based on"	No.	%
Last noted menstrual period	93	32.2
Date of conception	58	20.1
Date of procedure	6	2.1
Due date*	101	34.9
No response	31	10.7
Total	289	100.0

^{*} Correct response.

Table 20. Other Conditions Stated to Be Detectable by Maternal Serum AFP

No. of other conditions	No. of physicians	%
0	99	33.9
1	70	24.2
2	48	16.6
3	30	10.4
4	21	7.3
5	10	3.5
6	3	1.0
7	3	1.0
8	1	0.3
9	1	0.3
13	2	0.7
22	1	0.3
Total	289	100.0

Table 21. Physicians' Knowledge

	No.	%
Prenatal knowledge score		
0	42	14.5
1	158	54.7
2	89	30.8
Total	289	100.0

Table 21. (cont'd)

	No.	%
Maternal serum AFP knowledge score		
0	58	20.1
1	68	23.5
2	59	20.4
3	62	21.5
4	34	11.8
5	8	2.8
Total	289	100.0

Table 22. Quoted Sensitivity for Spina Bifida or Down Syndrome

	Spina bifida		Down s	yndrome
Sensitivity quoted	No.	%	No.	%
0-19	5	1.7	24	8.3
20-39	5	1.7	32	11.1
40-59	13	4.5	51	17.6
60-79	30	10.4	27	9.3
80-100	146	50.5	31	10.7
No response	90	31.1	124	42.9
Total	289	100.0	289	100.0

Note: 70-85 percent is correct response for spina bifida; approximately 35 percent is correct response for Down syndrome.

Table 23. Pattern of Physician Referral* for Prenatal Diagnosis

	No.	%
No response	21	7.3
Refer all women	213	73.7
Refer only certain women**	43	14.9
Never refer	12	4.2
Total	289	100.0

^{*} Referral means refers or offers referral.

^{**} Three refer for age other than stated cut-off, three refer for previous anomaly, and remainder did not specify which women are referred.

13

	No.	%
Maternal serum AFP testing is:		
No response	10	3.5
Done automatically on all patients as part of routine blood work	64	22.1
Done automatically on all patients unless patient specifically declines	56	19.4
Offered to all patients, done only if patient consents	109	37.7
Done for specific patients*	32	11.1
Not done at all	18	6.2
Total	289	100.0

rige grower man out on	10
Post-birth control pill conception	2
At patient's request	1
If patient very anxious	1
Reason not stated	5

Table 25.	Ways in Which Information Was	s Supplied Before
	Serum AFP Testing	

Previous anomaly or positive family history

Age greater than cut-off

Information supplied	No.	%
No response	27	9.3
None	19	6.6
Written only	4	1.4
Verbal only	155	53.6
Written and verbal	84	29.1
Total	289	100.0

Table 26 shows the significant associations found between various physician responses and characteristics. For the purpose of this table, significance was defined as a p value less than 0.01. The comparison group is shown in parentheses. For example, obstetricians were more likely than family or general practitioners to state that their patients decline AMA counselling because they would not abort. Some groups seem to be more knowledgeable than others regarding these issues. Not surprisingly, the average knowledge of obstetricians was greater than that of family or general practitioners in this area. Similarly, physicians who scored higher on prenatal diagnosis questions have practices with a higher percentage of obstetrics (mean percent obstetrics = 27.3 percent for 2 points vs. 11.0 percent for 1 point or 11.7 percent for 0 points) (p < 0.001). Likewise, those who scored higher on maternal serum AFP knowledge were the physicians who did more obstetrics (mean percent obstetrics = 52.1 percent for 5 points, 23.0 percent for 4 points, 21.8 percent for 3 points, 12.7 percent for 2 points, 11.3 percent for 1 point, 10.4 percent for 0 points) (p < 0.001).

In response to the question "How good a test is maternal serum AFP?" the scale was from 0 to 10, with 0 meaning the worst possible test and 10 meaning the best. The distribution of responses is shown in Figure 8.

Summaries of the physician comments are given in Tables 27 through 32. No specific pattern of maternal serum AFP use was found to be

associated with any specific type of comment.

In general, most physicians think that maternal serum AFP is a better than average test; the average response to the question "How good a test is maternal serum AFP?" was 6.3 on a scale from 0 to 10, but there is a wide range of opinions. Nine physicians thought the program should be cancelled, while six believed maternal serum AFP screening should be made mandatory. It is interesting to note that the view of physicians regarding how "good" a test maternal serum AFP is did not relate in any significant way to any of the physician characteristics except whether or not they refer all eligible women for AMA counselling. The mean rating for physicians who refer all of their eligible patients for AMA counselling was 6.4 compared to 4.0 for physicians who never refer for AMA counselling (p = 0.004). The views of physicians in this area did not seem to clearly relate to the pattern of the physicians' use of maternal serum AFP screening in their practice.

Table 26. Significant Positive Associations (Comparison Group Shown in Parentheses)

1. Physicians who stated "patients decline counselling because they wouldn't abort"

- Obstetricians (family/general practitioners)
- Physicians who supply written and/or verbal information re maternal serum AFP (physicians who do not supply information re maternal serum AFP testing)

2. Physicians who stated "false positives cause anxiety"

- 2 points for prenatal knowledge (< 2 points for prenatal knowledge)
- > 2 points for AFP knowledge (< 2 points for AFP knowledge)
- Obstetricians (family/general practitioners)
- Physicians who offer maternal serum AFP to all women (physicians who do not)

3. Obstetricians

- 2 points for prenatal knowledge (< 2 points for prenatal knowledge)
- > 2 points for AFP knowledge (< 2 points for AFP knowledge)
- Physicians who supply written and/or verbal information re maternal serum AFP (physicians who do not supply information re maternal serum AFP testing)

4. Female MDs

 Physicians who refer all eligible women for AMA counselling (physicians who do not)

5. First respondents

Physicians who offer maternal serum AFP to all women (physicians who do not)

6. Winnipeg physicians

- 2 points for prenatal knowledge (< 2 points for prenatal knowledge)
- > 2 points for AFP knowledge (< 2 points for AFP knowledge)
- Physicians who supply written and/or verbal information re maternal serum AFP (physicians who do not supply information re maternal serum AFP testing)
- Physicians who offer maternal serum AFP to all women (physicians who do not)

7. 2 points for prenatal knowledge

- Obstetricians or specialists (family/general practitioners)
- Winnipeg MDs (non-Winnipeg MDs)

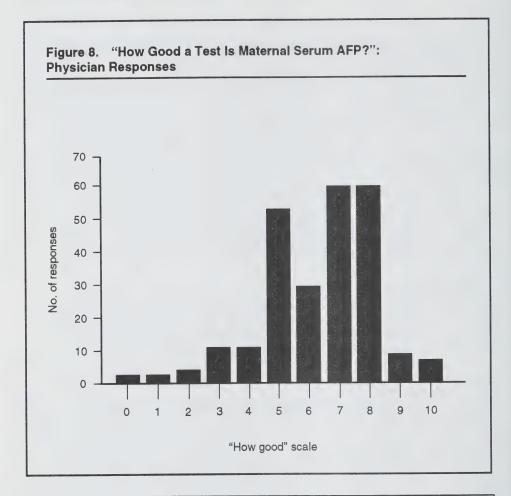


Table 27. Reason Patients Decline Counselling for AMA, According to Physicians

Reason	No.	%
Would not abort or would not change plans for pregnancy	110	38.1
Religious or moral reasons	36	12.5
Risk of procedure	33	11.4
"Do not want to know results"	16	5.5
Distance to Winnipeg	9	3.1
Personal reasons	4	1.4

Table 27. (cont'd)	Ta	ble	27.	(cont'd
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Reason	No.	%
History of infertility	2	0.7
First appointment too late for referral	2	0.7
Risk of abnormality considered low	2	0.7
First pregnancy	1	0.3
They believe everything will be okay	1	0.3
Risk of false positives	1	0.3
The patients do not like invasive tests	. 1	0.3
Their family history is negative	1	0.3
"They are from the Third World"	1	0.3

Table 28. Positive Aspects of Maternal Serum AFP Testing Seen by Physicians

Aspect	No.	%
Detection of anomalies	36	12.5
Reassuring if normal	36	12.5
Good screening test, accurate, effective, etc.	27	9.3
Simple or non-invasive test	18	6.2
Detection of anomaly and specifically mentions neural tube defects*	13	4.5
Prepares for birth of affected child (not necessarily for abortion)*	12	4.2
Detection of anomaly and specifically mentions abortion as option*	8	2.8
Detection of Down syndrome	5	1.7
Maternal serum AFP coordinator input helpful	3	1.0
Fetal assessment follow-up helpful	3	1.0

Table 28. (cont'd)

Physicians' comments

- 1. Gives patient choice if AFP positive.
- Good screening test, effective for parents wishing to use "new technology" to help prevent birth of a defective child.
- Abnormalities diagnosed allow patient to accept a diagnosis of Down syndrome before term (or of non-viable fetus or fetus with correctable defects).
- 4. It would be an important screening test where there is no ultrasound facility.
- Very helpful. If abnormal, the dates given by patient are re-checked and compared with ultrasound.
- 6. Comprehensive in approach to state-of-the-art obstetrics.
- 7. Like chicken soup, testing may help; not hurt.
- 8. May lead to a more careful fetal scanning than if not done.
- 9. Replaces need for routine scanning for anomalies.
- 10. Increases number of patients for genetic amniocentesis, therefore increases predictive value.
- Could alter their futures with more "choice" about family, preventing lifelong problems for parents if they are unwilling to cope with a "problematic" child.
- 12. Detects neural tube defects with high sensitivity to plan for Caesarian section and to prepare emotionally.
- Many knowledgeable primiparas worry endlessly re abnormalities so AFP helps reassure them.
- 14. Supplies useful information.
- * Subset of "detection of anomalies."

Table 29. Negative Aspects of Maternal Serum AFP Testing Seen by Physicians

Aspect	No.	%
Creates anxiety in patients and/or physicians	102	35.3
Too many false positives	68	23.5
Cost of screening or follow-up	26	9.0
Normal values can cause false reassurance	21	7.3
Leads to too many amniocenteses or invasive tests (i.e., increases risk)	18	6.2

Table 29. (cont'd)

Aspect	No.	%
Down syndrome detection part of program	16	5.5
Time or logistic constraints	10	3.5
Maternal serum AFP screening promotes abortion/abortion is wrong	8	2.8
Can lead to difficult decisions for patients	3	1.0
Report is too complex	2	0.7

Physicians' comments

- 1. It is overrated.
- 2. Most of the women I deal with would not want to know if the test is abnormal.
- Result can be confusing when gestational age does not equal age by dates.
- 4. In remote areas where investigations are delayed and expensive, this reduces benefit:cost ratio.
- 5. Patients need to be informed it does not ensure a normal baby.
- 6. Negative test does not ensure a perfectly normal pregnancy, so what is the point?
- 7. Follow-up fetal assessments not entirely reassuring.
- 8. Sometimes patient goes for nine months for fetal assessment but no abnormality is diagnosed. Waste of technician and resource.
- Something like 15 to 20 women are worried and subjected to unnecessary tests for each one picked up, and I do not know how to equate this with the benefit
- 10. It has failed to pick up twins for me (gestation was correct), I picked it up in the usual way but later than I would have otherwise.
- 11. Gives lots of unrealistic expectations re picking up Down syndrome (done too late in pregnancy to be considered a good screening test).
- 12. Late stage at which done makes result of questionable use if a reasonably non-traumatic abortion is planned after amniocentesis.
- 13. I have had four patients from relatively small numbers with false positive tests. This has caused much anxiety, frequent trips to Winnipeg, with risks of travel and considerable costs to patients.
- 14. It can put women in a position to make a decision they don't want to make.
- 15. Questionable whether should be done routinely.

Table 29. (cont'd)

Physicians' comments (cont'd)

- 16. I do not feel confident even if test is negative.
- 17. A number of my patients have been quite unsure of dates, hence repeated testing and potentially a lot of anxiety, especially with low values.
- 18. I have seen perhaps one low value in six years actually occurring where the infant born had Down syndrome.
- 19. Doesn't address moral/emotional dilemma.
- 20. Patients in this region are not particularly cognizant about spina bifida. Consequently, it is more to avoid the unpleasant work should this condition be present; "in a perfect society people demand a perfect product."
- 21. It is very complex to explain to mothers all the implications to truly achieve informed consent.
- 22. The medical-legal aspects are a concern.
- 23. Working outside Winnipeg, follow-up of abnormal results is difficult.
- 24. If you find a case of spina bifida or Down syndrome early in pregnancy, but it does not alter the course of the pregnancy (i.e., termination), there is very little benefit.

Table 30. Physicians' Suggestions for Improving Maternal Serum AFP Program

Suggestion	No.	%
More education for physician and/or patient	38	13.1
ncrease accuracy of test or find a more accurate test	9	3.1
Discontinue program	9	3.1
Improve turnaround time re Down syndrome detection	9	3.1
Make testing mandatory	6	2.1
Triple testing	4	1.4
Stop testing for Down syndrome	3	1.0
Make report simpler	3	1.0
Do ultrasound as a routine	3	1.0

Table 31. Other Physicians' Suggestions for Improving Maternal Serum AFP Program

Suggestions made

- Speed up turnaround time so that we're not doing karyotypes at 21 weeks.
- 2. The information card implies that the test is routine this should be changed. (I have to point out that the test is voluntary.)
- 3. Suggest scaling it down or replacing it with ultrasound screening.
- 4. Do not do the test as a routine without patient informed consent. (Verbal okay.)
- 5. Encourage use in those areas where ultrasound scan is not available. Do one follow-up scan and discharge at that juncture.
- Screen those "at risk" cases who would definitely proceed with abortion. Leave the rest alone.
- 7. Less follow-up in Winnipeg for false positives.
- 8. I plan to start recommending the program only to higher-risk patients to improve specificity.
- We are often requested to send ultrasound reports. It would be better (for the program) to phone and obtain relevant data.
- 10. Do not complicate the evaluation of this test with other various outcomes as that is not the reason we are testing, and other outcomes (intrauterine growth retardation, twins, etc.) can be determined by less cumbersome/ invasive means.
- 11. Do an evaluation looking at all costs/benefits of this program including emotions, stress in pregnancy/repeat tests/ultrasounds/amniocentesis, etc.
- 12. Develop another document that could be given to mothers for true informed consent.
- 13. I have a very, very busy practice. Could the program be expanded to "babysit" my patients by the coordinator rather than increase my workload (e.g., lengthy explanations of test results and recommendations)? Short and sweet information is efficient and means less work for me. I may be passing the medico-legal buck, but nowadays obstetricians need all the help they can get.
- 14. The maternal serum AFP program should have easy access to ultrasound reports so that offices do not need to phone in reports of ultrasound.
- 15. Treatment of affected fetuses rather than destruction.

Table 32. General Comments Received* from Physicians

- Use of percentage risk has very little meaning to the individual patient and there should be levels above which testing is not considered. The use of the level at which risks of test are less than the risk of abnormality as a justification for offering testing is causing a great deal of unnecessary fear and apprehension on the part of mothers who are already naturally worried about their infants. A mother who is told she has a 1 in 300 risk of Down syndrome only hears her baby has a higher chance of being abnormal.
- Vulnerable patients have been coerced into getting amniocentesis by wellmeaning but technologically oriented physicians because of this program. There should be safeguards against invasive procedures that carry risks to unborn babies.
- I resent the repeated intrusion of a second questionnaire rather than accepting my desire not to respond. I feel this questionnaire is directed to "improving" the service rather than establishing its morality or ethical standing. I do not accept the presumption that it is a "good idea."
- It is my interpretation that the only therapeutic intervention associated with this test is abortion. If as has recently been suggested an elevated AFP can be shown useful in screening for other high-risk pregnancies leading to delivery rather than termination then I would be prepared to become involved. I am ethically opposed to genetic screening for the purpose of aborting fetuses with genetic disorders such as Down syndrome or spina bifida.
- I have performed 40 to 60 deliveries annually for past four years. I've had no anomalous fetuses in patients with positive screens (average four to six positive screens per annum). This makes me seriously question the printed accuracy of the figures.
- In medical school we were trained not to miss the presence of treatable conditions. Neither of these (neural tube defects or Down syndrome) is a treatable condition, but abortion is an option. If abortion is not an option for the mother, then why spend time and money, not to mention risk of patient anxiety, with a false-positive test - when the management will not change anyway. I prefer to use ultrasound as the screen to rule out anencephaly.
- I highly resent therapeutic abortion being considered a "cure" for genetic disease/abnormality.
- The other problem with ante-natal screening is that it makes "elderly" mothers assume that any fetus allowed to go to term will be perfect which, of course, is not necessarily the case. It should be made plain that there are many problems that cannot be foreseen.
- I feel that a good ultrasound at 17 to 18 weeks provides the information regarding the fetus and other factors as well.

Table 32. (cont'd)

- Like most diagnostic technology, it has not been adequately tested in randomized control trials before its use and this should be done before implementation in other provinces.
- At this clinic we did do routine AFP as part of your research project some years ago. In the one case in the past 10 years where I have seen elevated AFP in a patient who was pregnant and over 30 years of age, she had ultrasound and eventually had a normal full-term pregnancy. Patient anxiety was a major problem.
- I don't believe in this, it is wrong. Why upset a woman if she doesn't plan to go ahead to have her baby killed; also, it may jeopardize a healthy baby, introduce infection, etc. We have to trust in God. Often the babies of older mothers (i.e., advanced maternal age) are very talented, very gifted and yet we see young mothers having Down syndrome babies, as in my own family. My relative's child has Down syndrome and they have had a healthy girl since and they wouldn't have amniocentesis. I have a cousin with spina bifida, a very healthy, happy young man in a wheelchair, married to a woman with severe osteogenesis imperfecta. Their 13-year-old son is a happy, healthy child. They are a happy, close family and very independent or as independent as most families are. They never discuss or complain regarding their handicaps.
- The other factors that limit what I can do in this population is that many come in too late for testing or their first exam is from 15 to 18 weeks or they have too many other pressing issues (alcoholism, etc.) to take time to explain the test. In the past, prior to the studies showing decreased disability with Caesarian in spina bifida infants, I asked women if they would have an abortion if the tests pointed to neural tube defects. If they said no, I would tell them not to have the test. Now with this study I leave it up to them.
- If it is "required" in all patients, we need to be told. At a lecture on the subject in 1991, several obstetricians were very derogatory in their remarks, leading one to believe that the test is a waste of time.
- I very strongly feel that this test should never be done as a routine part of prenatal blood work since the implications given an abnormal value are so serious. Parents should be given a free choice, without pressure, to decide if they want to have a screening test with full knowledge of what confirmatory tests would be available and for what purpose. In my experience most people decline the test. I am not in favour of this program. I feel there are too many ethical problems with obtaining this information if it leads to further tests and eventually to abortions of "abnormal" children.
- Present testing is inconvenient due to frequent requirement to have patient followed up earlier than normal pattern just to obtain AFP blood sample.

Table 32. (cont'd)

- If we are offering this test to confirm neural tube defects or Down syndrome with the intent that parents could terminate the pregnancy, then I see this as having serious ethical problems. Just because we have the technology to obtain this kind of information does not mean we should use it. We need to look at the ethical implications of this screening tool. Where is this kind of testing leading us? What other conditions will we be able to determine prenatally in the future? If there is no possibility to change the outcome for these children, then why should we access this information? It would seem to me that if our screening tests are ultimately leading to the abortion of "abnormal" children, then this can be compared with Hitler's Nazi campaign to kill off the disabled and "feeble minded" people of Germany at that time. If this is how we view the world as physicians (i.e., that disabled children should be considered for abortion) then this is a serious moral deficit in our thinking. [Note: this comment was in a letter co-signed by five physicians in one practice.]
- My knowledge of this is not 100% and I feel that I leave my patients without all information. However, I do feel I deal with this much better than many specialists do. I think this overall test should be looked at and its benefit to patients be re-evaluated.
- = The test needs to be clearly defined with good studies.
- I wonder whether this program would have been initiated if the results of the test were only valid between 20 and 25 weeks. This test does have benefits other than identifying pregnancies with the intention of termination for those with certain abnormalities. Would those other benefits have been sufficient to justify the program and make the test a part of standard practice in this province?
- Like most of the new technologies, it is hard to work out the ethics of benefit to a few at the price of much stress and worry for the many — I personally have no idea of the answer.
- I strongly feel that physicians need to discuss this test (maternal serum AFP) with patients and do it only on those who want it. The test has many moral/ethical implications, not just clinical/medical implications. However, most people will decide to have the test done.
- I am doing this test as a routine prenatal check-up. It is difficult to say how useful it is as so far I haven't had any patient with Down syndrome or spina bifida. If it is not expensive for health care it should be continued.
- Should continue to be an elective test for those who want to know the results because they would choose to alter the outcome.
- Have genetic counselling done as part of pre-marriage preparation. Pregnancy is not the time to do life or death investigations.

Table 32. (cont'd)

- Personally, I feel it is a good test but my patients haven't felt like they could deal with the results if abnormal given their options (i.e., a therapeutic abortion or waiting another four to five months for a deformed child instead of the "perfect" child everyone seems to hope for).
- + I think this is an excellent program.
- + I feel that Manitoba is fortunate to have a high-quality dedicated Human Genetics department.
- + Thank you for asking. I have been using this test routinely since long before it became a recommendation.
- + Important to continue with program.
- + Should extend program to other provinces. System is well-established in England and works very well.
- + An excellent resource. Thank you.
- + Working well.
- + Good program.
- + Comments received on form from lab are helpful.
- + Happy with program.
- + I think this is an excellent program.
- + Overall, I feel the program is excellent.
- + I appreciate your interest in doctors' opinions.
- + The AFP test picked up a complete anencephaly and spina bifida on a patient of mine who was 16 weeks pregnant. She really appreciated this early diagnosis.
- + I feel it is very important to have amniocentesis available for advanced maternal age. AFP should be available to all ladies regardless of age.
- Until last year I had some doubts about the usefulness of the test until one of my patients had an elevated AFP; she was found to have a fetus with a severe neural tube defect and was able to have an abortion at 19 weeks. Obviously, this is anecdotal but it has reinforced for me the reason for routine screening.

means primarily negative comments.

⁼ means primarily neutral comments.

⁺ means primarily positive comments.

The most strongly critical opinions about maternal serum AFP screening related to the issue of abortion. Some physicians think abortion is wrong under any circumstance; they believe that the program promotes abortion and therefore they are not in favour of maternal serum AFP screening. Many physicians commented that a negative aspect of maternal serum AFP screening is the increased anxiety caused by a high false-positive rate. This observation was made independent of the physicians' view toward maternal serum AFP screening. Increased patient anxiety after false-positive maternal serum AFP results has been documented by others (Abuelo et al. 1991; Keenan et al. 1991; Marteau et al. 1992). Abuelo et al. (1991) found that women with a low maternal serum AFP level tended to be more anxious than women seen for advanced maternal age even though the Down syndrome risk was similar. They suggested that one possible explanation may be the "surprise factor" of being informed of an abnormal result. In contrast, women seen for advanced maternal age often believed testing was routine.

Physicians are more in favour of the neural tube defect detection aspect of the program than the Down syndrome aspect; the negative view of maternal serum AFP screening for Down syndrome was not confined to

physicians who were against abortion.

Perhaps the most significant finding of this survey is the need for greater education of physicians and patients; this was the most frequent suggestion made for improving the maternal serum AFP program. Many physicians overestimated the abilities of maternal serum AFP screening, as shown by the overestimation of the sensitivity for Down syndrome. One physician commented that only one low value in six years of his practice was related to Down syndrome. Unless the physician had more than 120 patients in the six years with a low value (which would be unlikely), the positive predictive value in his practice would be equal to or better than expected. Overestimation of the capability of maternal serum AFP testing is also seen in several of the comments. For example, one physician said that the test failed to detect twins in his practice and the diagnosis was made later. Maternal serum AFP levels are elevated (2.5 MOM or greater) only in about 57 percent of cases of twins (Johnson et al. 1990). Consequently, a normal level should not decrease the clinical suspicion of Most physicians see only a limited number of patients with abnormal maternal serum AFP results; thus, very few see pregnancies with abnormal outcomes. One physician commented: "Until last year I had some doubts about the usefulness of the test until one of my patients had an elevated AFP; she was found to have a fetus with a severe neural tube defect and was able to have an abortion at 19 weeks. Obviously, this is anecdotal but it has reinforced for me the reason for routine screening."

Another interesting finding of this study relates to the differences between Winnipeg and non-Winnipeg physicians and patients. Winnipeg physicians stated fewer of their patients decline counselling for AMA compared to rural physicians. Nine of 123 physicians who practise more than 60 miles from Winnipeg stated that one of the reasons their patients decline AMA counselling is the distance to Winnipeg. Concerns were also raised about the cost and inconvenience of follow-up in Winnipeg, Non-Winnipeg physicians were less likely to perform maternal serum AFP testing routinely and consequently knew less about the test. Rural physicians were less likely to supply written or verbal information before testing compared to Winnipeg physicians. The non-Winnipeg physicians were also less informed about the test. This would appear to be a reflection of the greater access and use of maternal serum AFP screening in Winnipeg compared to the rest of the province. Distance to Winnipeg may be one factor, and efforts should be made to reduce this inequality of patient care. One possible improvement would be the development of prenatal outreach clinics. However, there are other reasons to explain the difference between Winnipeg and other Manitoba centres. Some of the non-Winnipeg centres may be more "conservative" and less likely to use prenatal testing. One of the most negative comments about maternal serum AFP came in a letter from a practice in a small, predominantly conservative town in eastern Manitoba. This practice of five doctors felt that the screening program should be discontinued as screening could lead to abortion of "abnormal" children

Summary

In many aspects the Manitoba Maternal Serum AFP Screening Program is functioning as anticipated. Its results are comparable to those obtained by other centres with respect to the sensitivity, false-positive rates, positive predictive value, and other screening parameters. However, it is clear that physician and patient education is one area where change is needed.

The birth prevalence of neural tube defects in Manitoba has declined markedly since 1979. For the six-year period from 1979 to 1984 the birth prevalence was 1.28 in 1 000 births, or approximately 21 infants per year, while from 1985 to 1990 the figure was 0.84 in 1 000 births, or 14 infants per year. When prenatal terminations are added to live births and stillbirths (1.38 in 1 000 births vs. 1.20 in 1 000 births) there is no observable decrease (Figure 6). Thus, this is due not to fewer occurring, but to fewer being live born. The total prevalence figures for Manitoba and the lack of trend over time show Manitoba has more similarities with continental Europe than with Britain in its patterns of neural tube defect incidence. In Manitoba, as in continental Europe, the decline that is apparent in neural tube defect prevalence has resulted almost entirely through prenatal diagnosis and termination of affected pregnancies. If most women found to be carrying an affected fetus continue to seek a termination of the pregnancy, the prevalence of these defects in term or

near-term infants should remain low, or even decline further, if the

proportion of screened women increases.

One further pertinent comparison can be made between our own and the European data. In Manitoba, as in Britain, a maternal serum AFP screening program is used by a high proportion of women. In such centres, terminations of pregnancies with neural tube defects occur at a much earlier stage of gestation than in continental Europe, where most prenatal diagnoses of neural tube defects are by ultrasound (EUROCAT Working Group 1991). Thus, in Paris and Strasbourg, for example, from 1984 to 1986, over 50 percent of induced abortions for spina bifida were done at gestations over 28 weeks and less than 10 percent under 20 weeks. In Wales, in comparison, where close to 50 percent of pregnant women were screened by maternal serum AFP, no induced abortions for spina bifida were done over 28 weeks, and 100 percent were under 20 weeks. Terminations for anencephaly tend to be somewhat earlier than those for spina bifida due to the ability of ultrasound to detect this defect earlier. In Manitoba in 1990, the average gestational age at termination of pregnancy for screened and detected fetuses with open neural tube defects was 19 weeks (range 17 to 22 weeks). However, for those cases detected initially by ultrasound, the average age at detection was 25 weeks (range 19 to 32 weeks). The European data led the EUROCAT Working Group (1991) to conclude that "... for spina bifida, the presence of a serum AFP screening program was linked to a much greater shift towards earlier diagnosis, indicating that in current routine practice conditions, the population sensitivity at earlier ages for spina bifida is greater than that of ultrasound screening." We would concur.

Some of the physicians surveyed suggested that maternal serum AFP screening could be replaced by ultrasound screening of all pregnant patients. Such a move should be treated with caution for several reasons. First, ultrasound is not appropriate as a screening test for Down syndrome. Second, in many regions, including both rural Manitoba and Winnipeg, either "routine" ultrasonographic examinations are not available or the waiting lists for appointments are several weeks long. Third, as the European data and our own suggest, reliance on ultrasound tends to lead to a delay in detection, giving the family fewer options when a major abnormality is detected. There are, therefore, strong reasons for a maternal

serum AFP program.

From our survey, Manitoba physicians generally are in favour of maternal serum AFP screening; however, some physicians are opposed. The major opposition is that prenatal screening promotes abortion. Physicians are more in favour of the neural tube defect detection aspect of maternal serum AFP testing than the Down syndrome detection aspect. Several factors likely account for this finding. Maternal serum AFP is about 70 percent to 85 percent sensitive for neural tube defects (Macrae et al. 1990) and, in 1990, for every 37 women with an elevated maternal serum AFP in our population, one was found to have a fetus with a neural tube

defect. More importantly, a fetal assessment can be done a few days after the woman is notified of high maternal serum AFP, and the fetal assessment usually can rule out a neural tube defect without the need for further tests.

Conversely, maternal serum AFP (and maternal age) is only about 35 percent sensitive for Down syndrome (Wald et al. 1988b). In Manitoba in 1990, only 1 in 110 women found to be at increased risk for having a baby with Down syndrome, but who had not already had an invasive prenatal test, did have such a fetus. To rule out Down syndrome in women with increased maternal serum AFP adjusted risks, an amniocentesis is necessary. The results of this test may not be available for four weeks, and the period of anxiety faced by the woman in this situation is therefore much greater. In addition, risks are associated with an amniocentesis, while a fetal assessment or ultrasound is considered to be a safe procedure. Many patients also worry that if the fetus is found to have Down syndrome, an abortion would not be an option given the relatively late gestation at which the diagnosis is made.

There is no doubt that maternal serum AFP screening for Down syndrome is more problematic than for neural tube defects. improvements could be made in this area. Nine physicians suggested a more accurate test be developed, while four others specifically suggested triple testing. Triple testing refers to the measurement of maternal serum beta-hCG and estriol in addition to maternal serum AFP to determine a woman's risk of having a child with Down syndrome. This would result in a much greater sensitivity and specificity. If an amniocentesis were done on the 5 percent of women at highest risk on the basis of their age (usually age 35 or older), the detection rate for Down syndrome would be approximately 20 percent. The average risk of Down syndrome would be about 1 in 140 (Knight et al. 1988). If an amniocentesis were done on the 5 percent of women at highest risk on the basis of their age and maternal serum AFP level, the detection rate would be 35 percent and the average risk would be about 1 in 120 (Cuckle et al. 1987). If an amniocentesis were done on the 5 percent of women at highest risk on the basis of their age and the triple test result, the detection rate would be 60 percent and the average risk would be 1 in 65 (Wald et al. 1988a). The introduction of triple testing has benefits for patients and physicians, but, even without triple testing, improvements can be made to help deal with patient anxiety.

Physician and patient education is an important issue in this regard. If women were counselled appropriately before an "abnormal" result is received, anxiety would likely be less; yet we found 6.6 percent of physicians do not supply any information before maternal serum AFP testing. We also found 22.1 percent of physicians do the test without getting specific consent. This is of concern, as women are then placed in a position of having knowledge and being presented with decisions they may not have wanted to consider. Many of the women found to be at increased risk for having a child with Down syndrome are already age 35

or older on their due date; offering early referral to these women (before maternal serum AFP testing) is appropriate but — given the results of the physicians' knowledge test — is not consistently done. The observed variations in practices of physicians are of concern. They mean that a woman's access to a funded service is determined by her doctor's attitudes, whereas it should be determined by her preferences, beliefs, and needs.

Several differences were documented between rural and Winnipeg practices in regard to prenatal diagnosis and maternal serum AFP testing. Some of the differences can be related to geographic factors and to differences in physician education and awareness. Attempts to reduce these inequalities should be made. Possible actions would include increased education about these issues for physicians, especially rural physicians. Prenatal outreach clinics may also be beneficial. Some of the differences in practice may also relate to more "conservative" views of

patients and physicians in rural settings.

We believe that the experience of the Maternal Serum AFP Screening Program in Manitoba indicates that such screening can be incorporated into standard prenatal practice in a coordinated and timely fashion. The impact of the program in terms of reduced birth prevalence of neural tube defects is clear and an effect on Down syndrome prevalence is starting to become apparent. Most patients and physicians support the availability of screening, though clearly it is important that it be offered in a way that leaves the decision whether or not to be screened to the individual patient. However, the data show that, in some cases, physicians' views concerning abortion may influence their decision whether or not to offer maternal serum AFP screening in their practices. It is our opinion that the views of all women should be respected and that no woman should have a prenatal test such as maternal serum AFP without it being her informed choice. We would recommend that all patients be offered testing and be free to decline if they wish.

Appendix 1. Patient Requisition

PRENATAL SERUM AFP

OBSTETRICAL HISTORY	
G P Rh .	
Comment:	
Hospital of Delivery	
FAMILY HISTORY	YES NO
Down syndrome	
Spina bifida/anencephaly	
Explain: (Name and relationship to patient)	
PRESENT PREGNANCY	YES NO
Chorionic villus sampling or amniocentesis	
Teratogen exposure	
Bleeding	
Explain:	
LNMP:	_
dd/mm/yy	
LNMP certain	-
Menses regular	
Every days	
Ultrasound	
If yes date of ultrasound Gestational age at U/S	
EDC	
	MP/US/Exam (circle one)
CURRENT INFORMATION	YES NO
Weightlbs/kg (circle one)	
Diabetic	
Multiple pregnancy	
Repeat sample	
	we routinely ask for four methods of gestational
age determination and check these	
Physician:	
Address:Phone:	
	7
Specimen preparation 1. Collect 10cc	Forward serum to Cadham Provincial Lab
2. Centrifuge	Metabolic Section
3. Forward serum only	750 William Avenue
4. Store at 4°C until shipping	Winnipeg, Manitoba R3C 3Y1
	naternal serum AFP screening (204) 788-6240.
	me of sample collection)
Draw date:	Gestational age at draw date:

Appendix 2. Patient Pamphlet

The AFP Blood Test

AFP stands for alpha-fetoprotein. It is made by your baby, and is present in your baby's blood and in the amniotic fluid around your baby. It is normal for some of this substance to reach the mother's bloodstream. Manitoba's AFP Screening Program was developed to provide mothers with the earliest information possible about normal development of their babies. If the exact length of pregnancy is known, we can tell which mothers have high, normal, or low amounts of AFP. Knowing the value for AFP can be very helpful in detecting problems that may affect your baby. In other words, we can do a simple test on the mother that helps us to learn about the fetus.

High AFP values are explained in many ways. Very often everything in the pregnancy is fine, even with a high value. Other explanations would be that the pregnancy is further along than suggested, there are twins, or the placenta (the afterbirth) is larger than expected. In all these cases, a normal baby or babies would be expected, although the test might be elevated. In some cases, birth defects such as spina bifida cause an elevation in the AFP — the AFP blood test was originally developed for this reason. Most twins, incorrect due dates, and babies with spina bifida are detected by high AFP results.

These complications of pregnancy are extremely important to your doctor in managing your pregnancy, and to you in preparing for the baby. A high AFP result may also suggest increased risk of other complications of pregnancy. These complications all involve problems in which the placenta does not work perfectly. These include problems of the baby's growth, high blood pressure in the mother, or premature labour. Such high-risk pregnancies are monitored more closely in the final months. If you have a high AFP result, you will have an ultrasound examination as soon as possible, to help discover the cause.

Low AFP values suggest another variety of pregnancy problems. The usual explanation is that the mother is not as far along in her pregnancy as she thought. In other situations, a low value may suggest problems with the baby's development. If you have a low result, you will receive detailed advice regarding the possibility of further investigations regarding your baby's health and development.

A normal result is very reassuring, to both you and your doctor. It means that severe spina bifida is very unlikely, and suggests a low risk of the other problems mentioned above. A normal AFP test does not, of course, guarantee a normal baby or a normal pregnancy. Your doctor uses the AFP test on a routine basis to help identify mothers at higher risk than usual. A single blood sample will be taken from you between the 15th and 18th weeks of pregnancy.

Your doctor routinely offers this test, and your doctor will contact you if further tests are required after the first sample.

Your doctor will continue to manage your pregnancy with careful attention to all details, even if you have a normal AFP result. If you do not wish to have this test please notify your doctor.

This card is for general information about the test. If you have an abnormal result on your AFP test, you will receive much more detailed information. An abnormal test does not prove there is a problem, but it means further tests should be done. If your blood test shows an abnormality, you will be contacted by your doctor, and further investigations will be organized for you.

This program is funded by the Province of Manitoba, and is run from a central office. Please contact us if you require further information.

Appendix 3. Sample Patient Report Form

MATERNAL SERUM AFP SCREENING PROGRAM

PRESENT PREGNANCY:
LNMP: 20-Dec-91 FREQ: 28 days
SURE: EDC supplied: 27-Sep-92
U/S DATE: GEST. ON U/S wks Age ON DUE DATE: 39 years
DATE SAMPLE DRAWN: 16-Apr-92 GESTATION (supplied): 16.5 wks. GESTATION (calculated and used in interpretation): 16.5 wks.
TWINS: No NEURAL TUBE DEFECT: No DOWN SYNDROME: No BLEED: Yes Amniocentesis or Chorionic Villus Sampling done: No
RESULTS:
AFP: 89µg/L Multiples of Median (MOM): 2.20
(Normal range = $0.45 - 2.2 \text{ MOM}$)
**** Down syndrome risk: 1 in 146 or equal to a 39-yrold ***** Correction used for weight: No (59kg), for diabetes: No

THE AFP IS NORMAL. However, as the patient will be over age 35 by the due date an amniocentesis may be offered. As the Down syndrome risk is not increased by this AFP level, the quoted risk is based on age alone.

If amniocentesis desired, call 787-4804.

F

INTERPRETATION:

- If an ultrasound is to be (or has been) done between 15 and 20 weeks' gestation, please send us a copy of the report.
- * If significant bleeding has occurred in last four weeks, please send a sample at the next appointment.

PLEASE CHECK THAT ALL INFORMATION, ESPECIALLY THE GESTATION, IS ACCURATE.

PLEASE NOTIFY US IF THERE ARE DISCREPANCIES.

Appendix 4. Survey Questionnaire

Physician Questionnaire

1.	What percentage of your practice directly relates to obstetrics? If the answer to this question is 0%, please simply return the questionnaire with the remaining questions unanswered. Please provide any comments you wish on this subject.
2.	PERSONAL INFORMATION Year of birth: place of birth:
	Graduation from medical school: year university
	I am a (circle best answer) (a) family or general practitioner (b) obstetrician/gynaecologist (c) subspecialist obstetrician/gynaecologist (specify):
	I practice (a) in Winnipeg (b) within 60 miles of Winnipeg (c) beyond 60 miles of Winnipeg
	Religion: Sex:
3.	PRENATAL DIAGNOSIS FOR ADVANCE MATERNAL AGE (a) At what age are women eligible for an amniocentesis or chorionic villus sampling based on their age alone? years

he date of the procedure he due date
arding the women who are eligible based on their age, I routinely refer all women only refer certain patients (please explain) never refer for this indication
ent:
roportion of your referred patients decline genetic counselling ning advanced maternal age? s for declining:
RNAL SERUM ALPHA-FETOPROTEIN (AFP) TESTING v sensitive (as a percentage) is maternal serum AFP for spina bifida? Down syndrome?
at other conditions may be detected by maternal serum AFF ing?
ternal serum AFP testing is done automatically on all my patients as part of their routine prenatal blood work done automatically on all my patients unless patient

604 Current Practice of PND

Comment:	
(d) What information is given to patients in advance of testing? (i) none (ii) written hand-out only (iii) verbal explanation only (iv) both written and verbal	
Comment:	
(e) How much time is spent discussing the maternal serum AFP with each patient? minutes	test
(f) How "good" a test do you feel maternal serum AFP is? (0 = w possible test, 10 is best possible test)	orst
(g) What do you feel are the positive and/or negative aspect maternal serum AFP testing?	s of
(h) What suggestions do you have for improving the program?	

Appendix 5. Comments Made by Physicians Who Do Not Practise Obstetrics

I provide primary care for mentally retarded people. More than 50% are due to unknown causes. I agree with all new prenatal diagnostic techniques that can help prevent the birth of a disabled child or a mentally retarded person.

I'm glad someone is wasting my money on this survey. Would agree with program.

Too bad the test was not available in my era as I have delivered babies with both spina bifida and Down syndrome.

My practice did have a severe septic abortion following a 16th week amniocentesis. Even though this may have been rare, it was very frightening.

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Mandate

(approved by Her Excellency the Governor General on the 25th day of October, 1989)

The Committee of the Privy Council, on the recommendation of the Prime Minister, advise that a Commission do issue under Part I of the Inquiries Act and under the Great Seal of Canada appointing The Royal Commission on New Reproductive Technologies to inquire into and report on current and potential medical and scientific developments related to new reproductive technologies, considering in particular their social, ethical, health, research, legal and economic implications and the public interest, recommending what policies and safeguards should be applied, and examining in particular,

- (a) implications of new reproductive technologies for women's reproductive health and well-being;
- (b) the causes, treatment and prevention of male and female infertility;
- (c) reversals of sterilization procedures, artificial insemination, in vitro fertilization, embryo transfers, prenatal screening and diagnostic techniques, genetic manipulation and therapeutic interventions to correct genetic anomalies, sex selection techniques, embryo experimentation and fetal tissue transplants;
- social and legal arrangements, such as surrogate childbearing, judicial interventions during gestation and birth, and "ownership" of ova, sperm, embryos and fetal tissue;
- (e) the status and rights of people using or contributing to reproductive services, such as access to procedures, "rights" to parenthood, informed consent, status of gamete donors and confidentiality, and the impact of these services on all concerned parties, particularly the children; and
- (f) the economic ramifications of these technologies, such as the commercial marketing of ova, sperm and embryos, the application of patent law, and the funding of research and procedures including infertility treatment.

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